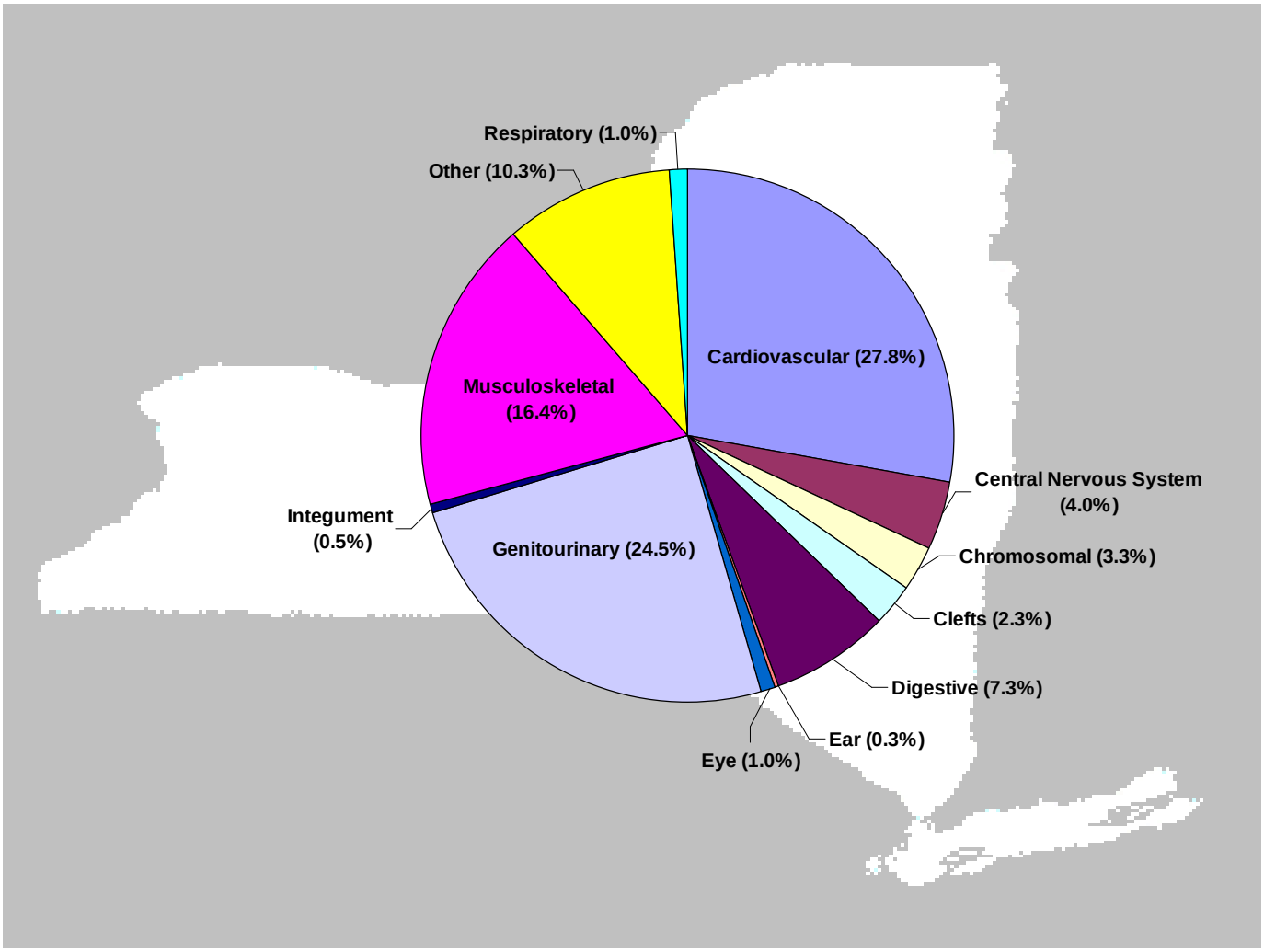
New York State Department of Health

Congenital Malformations Registry Summary Report



Statistical Summary of Children Born in 2005 and Diagnosed Through 2007

Additional and related information is also available from the New York State Department of Health Web site on the Internet: http://www.health.state.ny.us

Comments regarding the format or content of this report are welcome.

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Summary

This Congenital Malformations Registry Summary Report presents rates of congenital malformations occurring among the 242,659 children who were born alive to New York residents in 2005. The children reported with a major congenital malformation represent 4.8 percent of live births. Males had a higher rate of major congenital malformations than females (5.8 percent versus 3.8 percent), and black children had a higher major malformation rate than white children (6.0 percent versus 4.7 percent). This information is provided through mandated reporting by hospitals and physicians.

Demographic characteristics of those children reported to the Congenital Malformations Registry (CMR) and number of malformations are included in the report. Other sections present the distribution of anomalies by organ system; rates for selected malformations by race and sex and the most common malformations for each county are also included.

This is the seventeenth report from the CMR. Reports are also available by request for the 1983 to 2004 birth cohorts. This report and the reports for 1994-2004 are also available on the Department of Health website. The statistics in this report are **not** comparable to reports before 1992. In 1992, the CMR began to use a new coding system that allows for greater detail in coding. For previous years, ICD-9 codes were used. Information from birth certificates was used to supplement or correct reported data. Birth certificate matching also helps eliminate duplicate cases reported under different names and nonresident births. Reports produced for 1989 to 1991 did not use birth certificate matching.

PROGRAM OVERVIEW

Background

Congenital malformations are the leading cause of infant mortality in the United States.[1] They are the fifth leading cause of years of potential life lost and a major cause of morbidity and mortality throughout childhood.[1,2] Twenty percent of infant deaths are attributed to congenital malformations,[2] a percentage that has increased over time.[1,2] Approximately 25 percent of pediatric hospital admissions and about one-third of the total number of pediatric hospital days are for congenital malformations of various types.[3] Little is known about the causes of congenital malformations. Twenty percent may be due to a combination of heredity and other factors; 7.5 percent may be due to single gene mutations; 6 percent to chromosome abnormalities; and 5 percent to maternal illnesses, such as diabetes, infections or anticonvulsant drugs.[4] Approximately 40 percent to 60 percent of congenital malformations are of unknown origin.[4,5]

Although radiation and rubella had been linked to birth defects, not until the thalidomide tragedy of the early 1960s was there a widespread interest in possible associations between congenital malformations and environmental agents. During the 1970s, interest continued to grow in birth defects and birth defects surveillance as a result of the growing recognition of the problems of toxic waste dumps such as Love Canal and accidents such as Three Mile Island and Seveso. In response, many states began to develop birth defects registries in order to have data for tracking trends in malformation rates.[6,7] A birth defects registry also makes it possible to respond to public concerns about possible excess occurrence of malformations with timely, objective investigations. A birth defects registry can provide cases for traditional epidemiologic studies of specific congenital malformations and provide information for the planning, provision and evaluation of health services.[6,7]

New York State
Congenital Malformations Registry

The New York State Department of Health Congenital Malformations Registry (CMR) is one of the largest statewide, population-based birth defects registries in the nation. The concept of the Congenital Malformations Registry arose out of recognition of the environment as a potential etiologic factor in the occurrence of congenital malformations. Health studies during the Love Canal crisis in 1978 to 1983 confirmed the inadequacies of relying on birth certificates to monitor and evaluate birth defects.

New York's Congenital Malformations Registry was established by enactment of Part 22 of the State Sanitary Code in 1981. Reporting to the registry began in October 1982. Hospitals and physicians are required to report children under two years of age diagnosed with a malformation. The majority of reports are sent by hospitals, primarily from their medical records departments. A small number are sent by individual physicians to verify diagnoses initially suspected in the hospital but confirmed on an outpatient basis, and to clarify nonspecific diagnoses reported by hospitals.

The Congenital Malformations Registry receives case reports on children diagnosed up to two years of age who were born or reside in New York State with a congenital malformation, chromosomal anomaly or persistent metabolic defect. For purposes of this registry and report, a congenital malformation is defined as any structural, functional or biochemical abnormality,

determined genetically or induced during gestation and not due to birthing events.

Case reports are received electronically on the Internet using the Health Provider Network (HPN). The Department of Health developed the HPN as a secure system for electronically collecting and distributing health-related data. Pertinent fields are coded and the narrative description of the malformation is converted to a code. The case report is matched to existing registry reports for possible duplicates. Data submitted on HPN using either online data entry forms or file upload facility are transferred to a DOH UNIX server for updating of the CMR database.

All information reported to the registry is held in strict confidence. Records and computer files are maintained in accordance with DOH regulations concerning data containing individual identifiers. Access to the data by anyone other than registry personnel is restricted and carefully monitored to ensure that confidentiality is maintained. Families of children reported to the registry are never contacted without prior consent of the DOH's Institutional Review Board and notification of the child's physician.

2005 Report

This current report presents statistics for major anomalies only (see Appendix 1 and the glossary of birth defects in Appendix 5). This is in accordance with the practices of other state birth defects registries and allows comparison between New York State rates and rates in other states. Minor anomalies may cause problems in the determination of malformation rates because they are common and variably reported. They may not even be recorded in the medical chart.

The statistics in this report are **not** comparable to reports prior to 1992. The 2005 report is based on birth certificate matched cases (Appendix 2) with resident live births from the vital records file used as the denominator. The available birth certificate fields are used to supplement or correct reported data. Birth certificate data are used to establish maternal residence at birth. Birth certificate matching helps eliminate duplicate cases reported under different names. Racial data are not comparable because race is defined by maternal race from the birth certificate. Using maternal race is a common practice among birth defects registries nationwide as the race of the father is poorly reported. In earlier years, race was defined by what was reported on the CMR form, which may differ from what is recorded on the birth certificate. In 1992, the registry began using a new coding system, the modified British Pediatric Association code (BPA). This coding scheme is used by a number of other congenital malformations registries and allows for greater specificity than does the ICD-9 system. Since 1992, the list of major malformations has been revised (see Appendix 4) changing the list of major malformations used in Sections I and II and the number of specific malformation prevalences in Section III.

CMR Birth Cohort reports are intended as a resource for programs providing primary, secondary and tertiary preventive health care and for public officials concerned with reducing overall mortality and morbidity. The first annual cohort included children born in 1983 and reported with a malformation diagnosed before their second birthday.[9] This report describes children born in 2005 and diagnosed before their second birthday. Reports are also available for the 1984 through 2004 birth cohorts. Some reports and additional information are available through the DOH Web site at http://www.health.state.ny.us/diseases/congenital_malformations/cmrhome.htm.

Limitations

Care should be taken in the use of these data. Virtually all reports are abstracted from inpatient hospital records, since malformations diagnosed on an outpatient basis are not well reported. Accurate hospital clinical recognition of malformations depends on clinical acumen and interest. This is particularly true of conditions more difficult to diagnose, such as fetal alcohol syndrome. Consequently, identification of malformations may vary by area and by time. The abstracting of records requires well-trained medical records professionals who are fastidious in their reporting of such findings. Areas with hospitals that provide higher levels of care may have more thorough diagnoses and, thus, apparently higher rates. Similarly, areas with hospitals that report cases more completely will also appear to have higher rates. In regions with low numbers of births, small variations in incidence may produce large statistical fluctuations.

New York State Population

Based on the U.S. 2000 census, the population of New York State was about 19.0 million; more than 42 percent of the population lived in New York City. An additional 23 percent of the population lived in the six counties closest to New York City. In 2005, there were 242,659 resident live births reported to the state's vital registration, 16.9 percent to black mothers, and 23.3 percent to Hispanic mothers. In accordance with the practices of other state birth defects registries, the race of the child is based on race of the mother only. Nearly 48.2 percent of live births were to New York City residents.

References

1. Kochanek KD, Hudson BC. Advanced report of final mortality statistics, 1992. *Monthly Vital Statistics Report* 1995; 43(6 suppl.). Hyattsville (MD):National Center for Health Statistics, 1995.

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8. Merlob P, Papier CM, Klingberg MA, Reisner SH. Incidence of congenital malformations in the newborn, particularly minor abnormalities. In: Marois, ed. *Prevention of physical and mental congenital defects, Part C: Basic and medical sciences, education and future strategies. Proceedings of a conference of the Institut de la Vie*. New York: Alan R. Liss, 1985: 51-53.

9. New York State Department of Health. *Congenital Malformations Registry Annual Report: 1983 Birth Cohort.*

Section I
Demographic Characteristics of Children Reported with Major Malformations

Introduction to Tables

These tables are based on children resident in New York State who were live born in 2005 and reported to the registry with major malformations. Since a new coding system began to be used in 1992, the list of major malformations has been revised (see Appendix 4). Thus, the prevalence in this report are not comparable to reports prior to 1992.

The overall occurrence of major malformations was 4.8 percent of live births. Male children have a higher rate of major malformations than female children (5.8 percent versus 3.8 percent, Table 1). This difference is consistent within different racial groups. The rates for major malformations are somewhat higher for black than for white children (6.0 percent versus 4.7 percent). The major malformation rate among children with residence at birth in New York State excluding New York City was comparable to that among children with residence at birth in New York City (4.8 percent versus 4.8 percent). The smaller number of births in the "other" racial category makes these rates difficult to interpret.

About 82 percent of children reported with major malformations have only one major malformation (Table 2). Since most children had one major malformation, the race-sex patterns seen for all major malformations are similar to the patterns seen in children with a single major malformation (Table 3). All race-sex groups for children with multiple major malformations showed little variation (Table 4).

Section 1 - Table 1
2005 Births - New York State Residents
Percent of Live Births with One or More Major Malformations
Sex by Race/Ethnicity and Residence

	Both Sexes			Males			Females		
Race and Residence	Infants	Total Births	%	Infants	Total Births	%	Infants	Total Births	%
New York State									
- All Races	11,676	242,659	4.8	7,228	124,392	5.8	4,448	118,267	3.8
- Non-Hispanic White	5,699	121,416	4.7	3,623	62,184	5.8	2,076	59,232	3.5
- Non-Hispanic Black	2,442	40,772	6.0	1,430	20,857	6.9	1,012	19,915	5.1
- Hispanic	2,589	56,558	4.6	1,557	28,927	5.4	1,032	27,631	3.7
- Others/Unknown	946	23,913	4.0	618	12,424	5.0	328	11,489	2.9
NYS Excluding NYC									
- All Races	6,076	125,659	4.8	3,865	64,454	6.0	2,211	61,205	3.6
- Non-Hispanic White	4,250	88,538	4.8	2,724	45,323	6.0	1,526	43,215	3.5
- Non-Hispanic Black	705	12,042	5.9	420	6,087	6.9	285	5,955	4.8
- Hispanic	827	18,054	4.6	514	9,346	5.5	313	8,708	3.6
- Others/Unknown	294	7,025	4.2	207	3,698	5.6	87	3,327	2.6
New York City									
- All Races	5,600	117,000	4.8	3,363	59,938	5.6	2,237	57,062	3.9
- Non-Hispanic White	1,449	32,878	4.4	899	16,861	5.3	550	16,017	3.4
- Non-Hispanic Black	1,737	28,730	6.0	1,010	14,770	6.8	727	13,960	5.2
- Hispanic	1,762	38,504	4.6	1,043	19,581	5.3	719	18,923	3.8
- Others/Unknown	652	16,888	3.9	411	8,726	4.7	241	8,162	3.0

Section 1 - Table 2
2005 Births - New York State Residents
Number of Major Malformations Per Child

Number of Malformations	Number of Children	Percent
1	9,555	81.8
2	1,442	12.4
3	416	3.6
4	149	1.3
5	70	0.6
6	27	0.2
7	8	0.1
8	4	*
9	3	*
10	2	*
All Children	11,676	100.0

* - Less than 0.05%

Note: Total percent may not add to 100% due to rounding

Section 1 - Table 3
2005 Births - New York State Residents
Percent of Live Births with One Major Malformation
Sex by Race/Ethnicity and Residence

	Both Sexes			Males			Females		
Race and Residence	Infants	Total Births	%	Infants	Total Births	%	Infants	Total Births	%
New York State									
- All Races	9,555	242,659	3.9	6,002	124,392	4.8	3,553	118,267	3.0
- Non-Hispanic White	4,649	121,416	3.8	3,018	62,184	4.9	1,631	59,232	2.8
- Non-Hispanic Black	2,000	40,772	4.9	1,186	20,857	5.7	814	19,915	4.1
- Hispanic	2,132	56,558	3.8	1,287	28,927	4.4	845	27,631	3.1
- Others/Unknown	774	23,913	3.2	511	12,424	4.1	263	11,489	2.3
NYS Excluding NYC									
- All Races	4,943	125,659	3.9	3,213	64,454	5.0	1,730	61,205	2.8
- Non-Hispanic White	3,442	88,538	3.9	2,259	45,323	5.0	1,183	43,215	2.7
- Non-Hispanic Black	581	12,042	4.8	355	6,087	5.8	226	5,955	3.8
- Hispanic	679	18,054	3.8	434	9,346	4.6	245	8,708	2.8
- Others/Unknown	241	7,025	3.4	165	3,698	4.5	76	3,327	2.3
New York City									
- All Races	4,612	117,000	3.9	2,789	59,938	4.7	1,823	57,062	3.2
- Non-Hispanic White	1,207	32,878	3.7	759	16,861	4.5	448	16,017	2.8
- Non-Hispanic Black	1,419	28,730	4.9	831	14,770	5.6	588	13,960	4.2
- Hispanic	1,453	38,504	3.8	853	19,581	4.4	600	18,923	3.2
- Others/Unknown	533	16,888	3.2	346	8,726	4.0	187	8,162	2.3

Section 1 - Table 4
2005 Births - New York State Residents
Percent of Live Births with Two or More Major Malformations
Sex by Race/Ethnicity and Residence

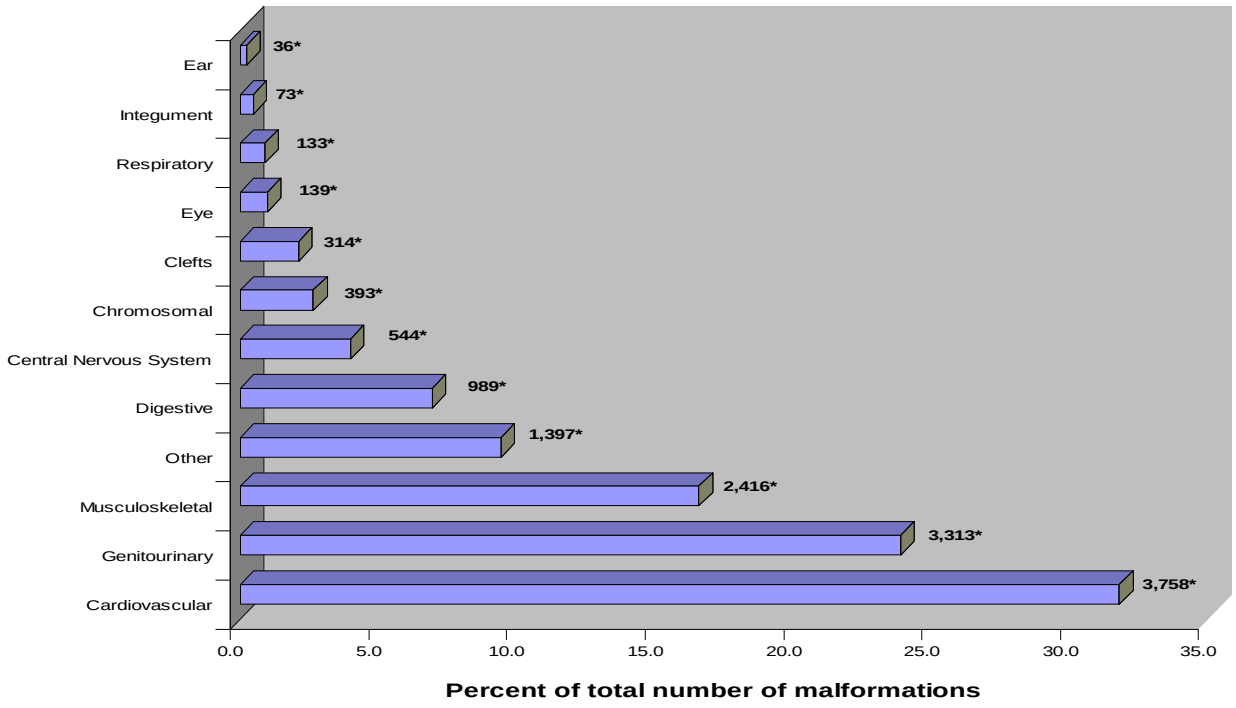
	Both Sexes			Males			Females		
Race and Residence	Infants	Total Births	%	Infants	Total Births	%	Infants	Total Births	%
New York State									
- All Races	2,121	242,659	0.9	1,226	124,392	1.0	895	118,267	0.8
- Non-Hispanic White	1,050	121,416	0.9	605	62,184	1.0	445	59,232	0.8
- Non-Hispanic Black	442	40,772	1.1	244	20,857	1.2	198	19,915	1.0
- Hispanic	457	56,558	0.8	270	28,927	0.9	187	27,631	0.7
- Others/Unknown	172	23,913	0.7	107	12,424	0.9	65	11,489	0.6
NYS Excluding NYC									
- All Races	1,133	125,659	0.9	652	64,454	1.0	481	61,205	0.8
- Non-Hispanic White	808	88,538	0.9	465	45,323	1.0	343	43,215	0.8
- Non-Hispanic Black	124	12,042	1.0	65	6,087	1.1	59	5,955	1.0
- Hispanic	148	18,054	0.8	80	9,346	0.9	68	8,708	0.8
- Others/Unknown	53	7,025	0.8	42	3,698	1.1	11	3,327	0.3
New York City									
- All Races	988	117,000	0.8	574	59,938	1.0	414	57,062	0.7
- Non-Hispanic White	242	32,878	0.7	140	16,861	0.8	102	16,017	0.6
- Non-Hispanic Black	318	28,730	1.1	179	14,770	1.2	139	13,960	1.0
- Hispanic	309	38,504	0.8	190	19,581	1.0	119	18,923	0.6
- Others/Unknown	119	16,888	0.7	65	8,726	0.7	54	8,162	0.7

Section II
Major Congenital Malformations by Organ System, 2005

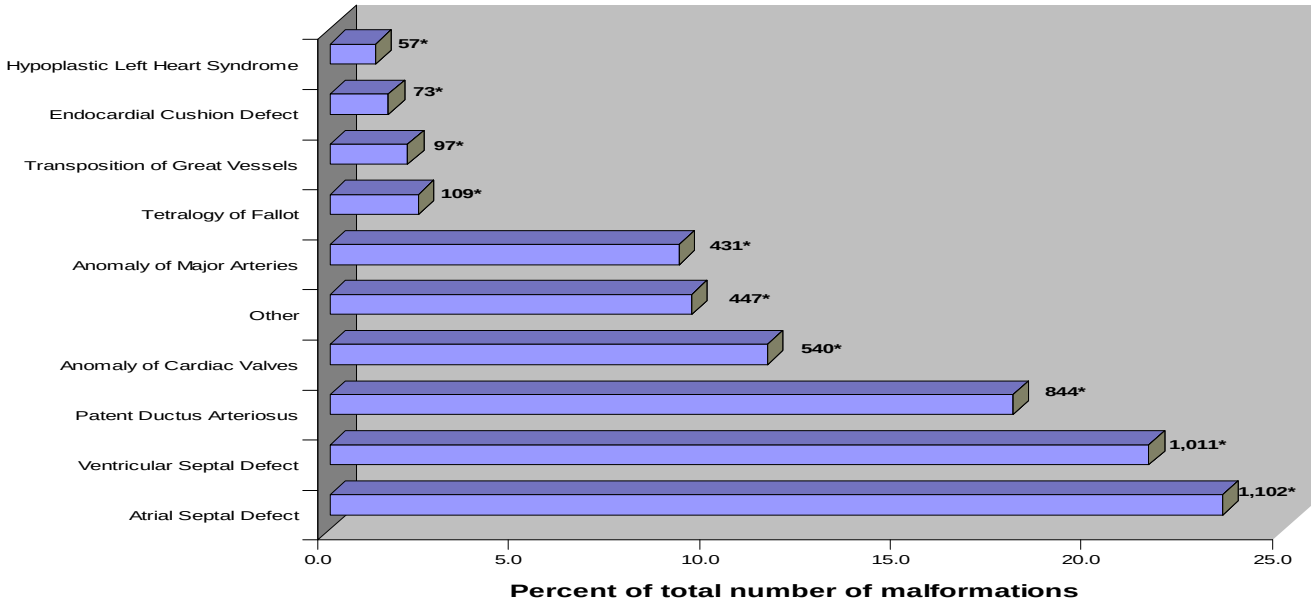
Introduction to Figures

The organ system figures in this section present the distribution of 12 categories of major malformations, the relative contribution of each category to overall prevalence of major malformations in New York State, and the contribution of type of malformation within each subset category. Some of these percentages may differ from previous reports because of the new malformation coding system described in the program overview.

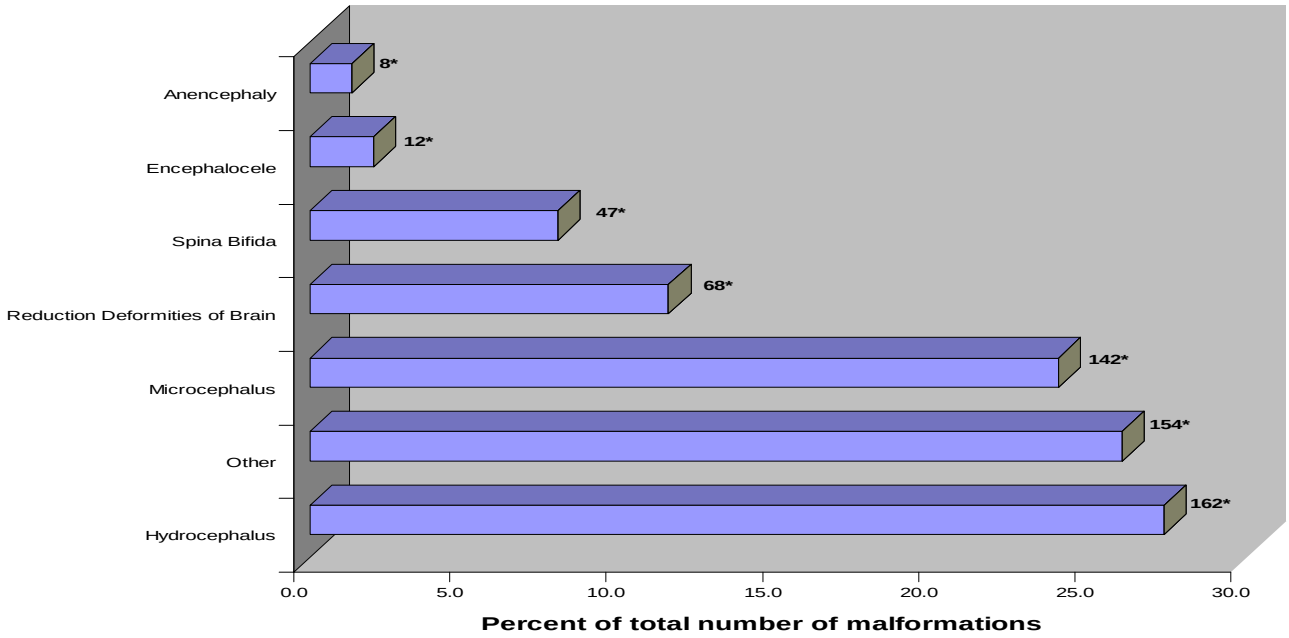
Major Malformations by Organ System
2005 Births - New York State Residents
(Number of Children = 11,676)
(* - Number of malformations in each organ system)



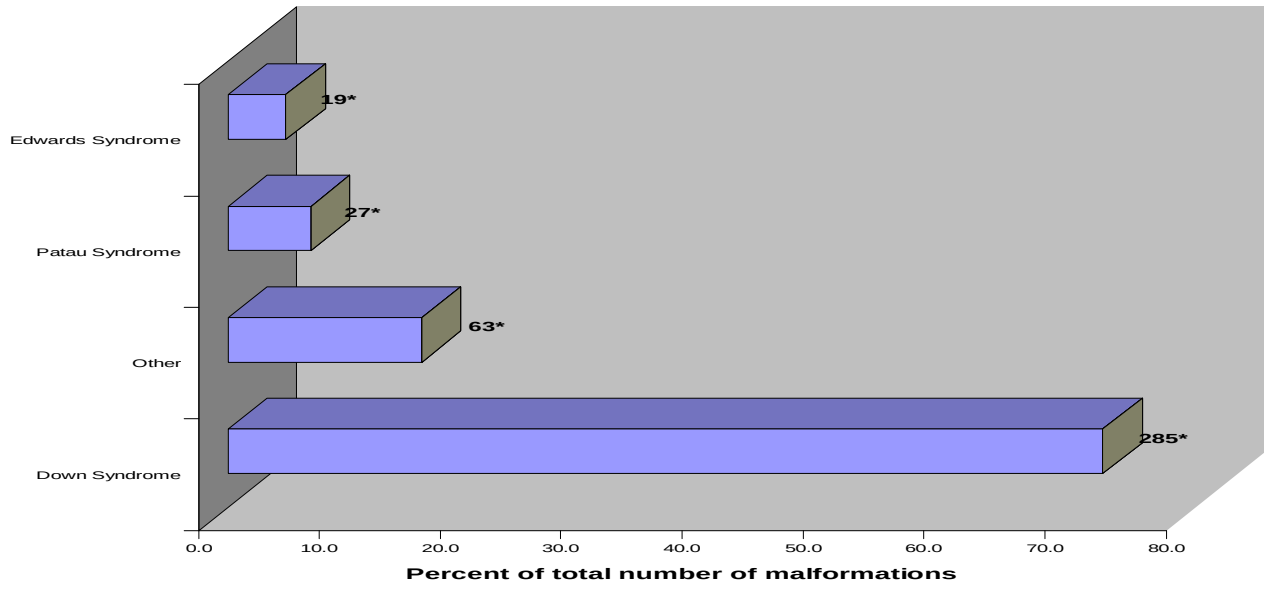
Major Malformations by Organ System
2005 Births - New York State Residents
Cardiovascular System Subset Category
(Number of Children = 3,758)
(* - Number of malformations in each category)

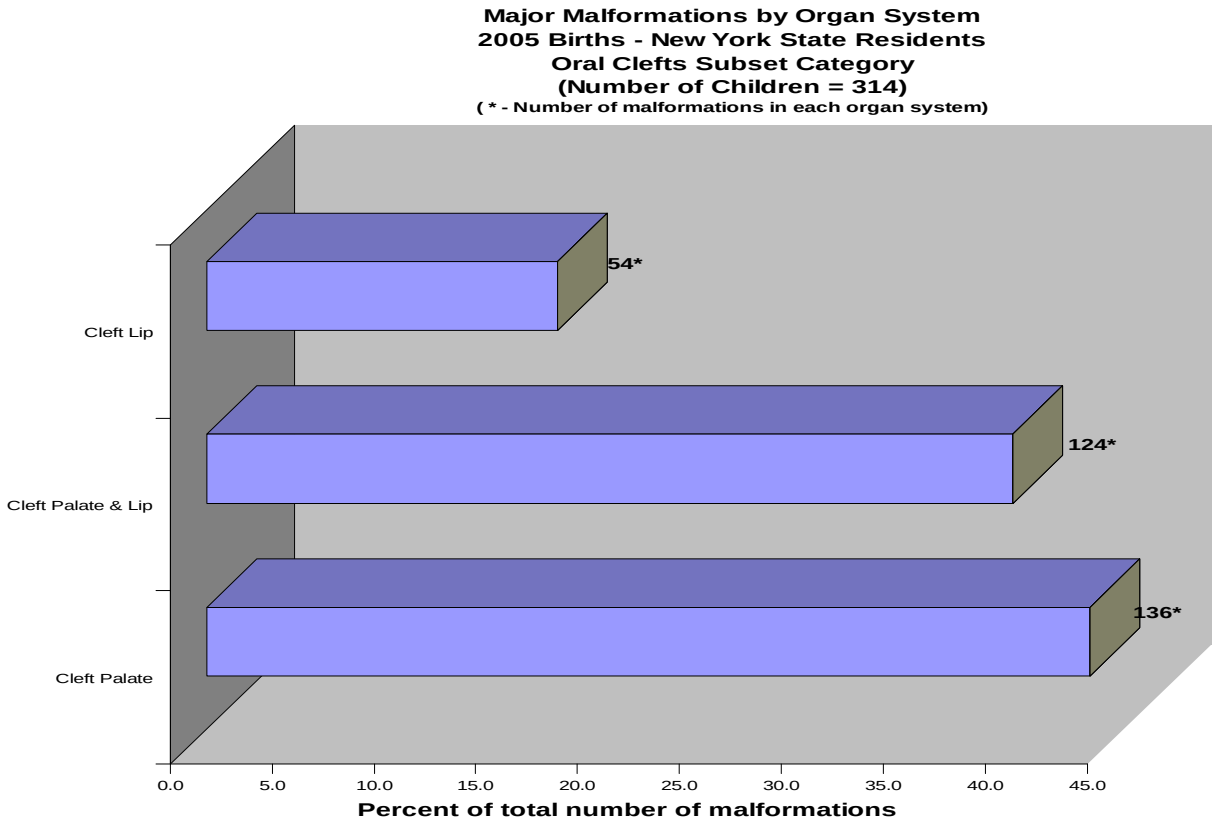


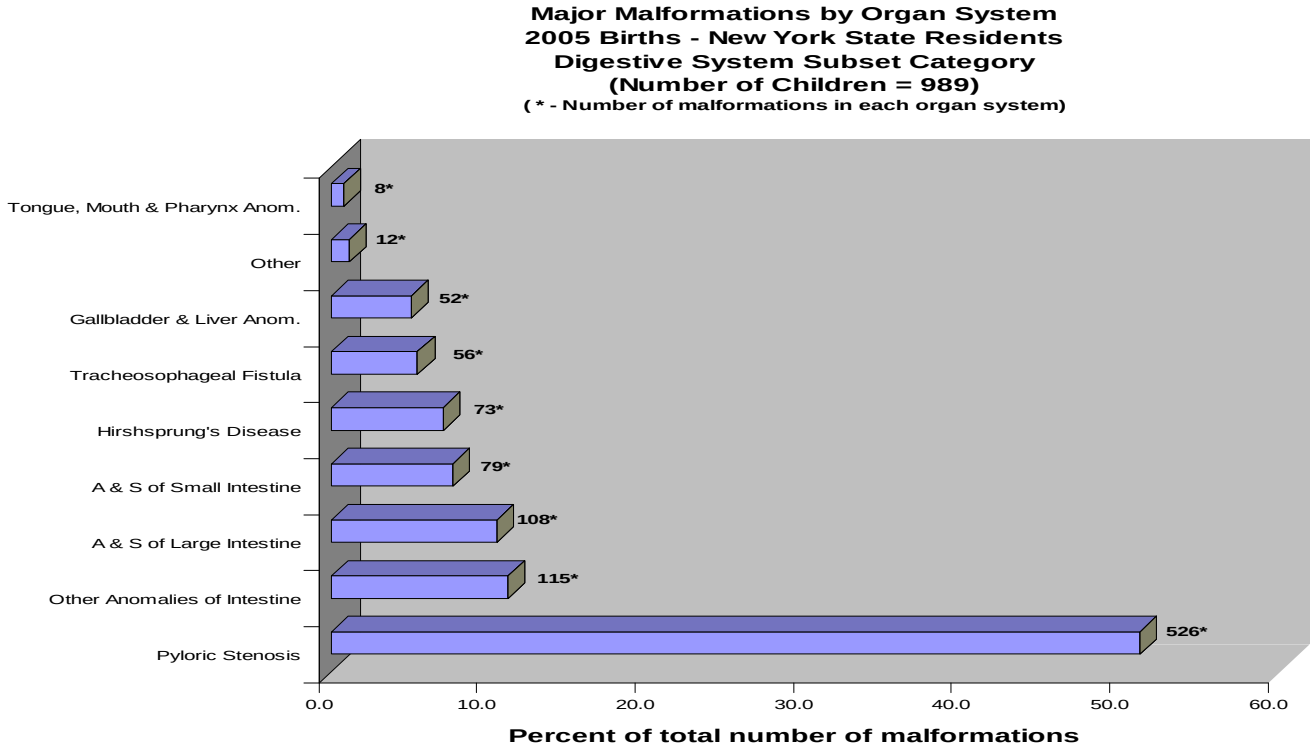
Major Malformations by Organ System
2005 Births - New York State Residents
Central Nervous System Subset Category
(Number of Children = 544)
(* - Number of malformations in each organ system)



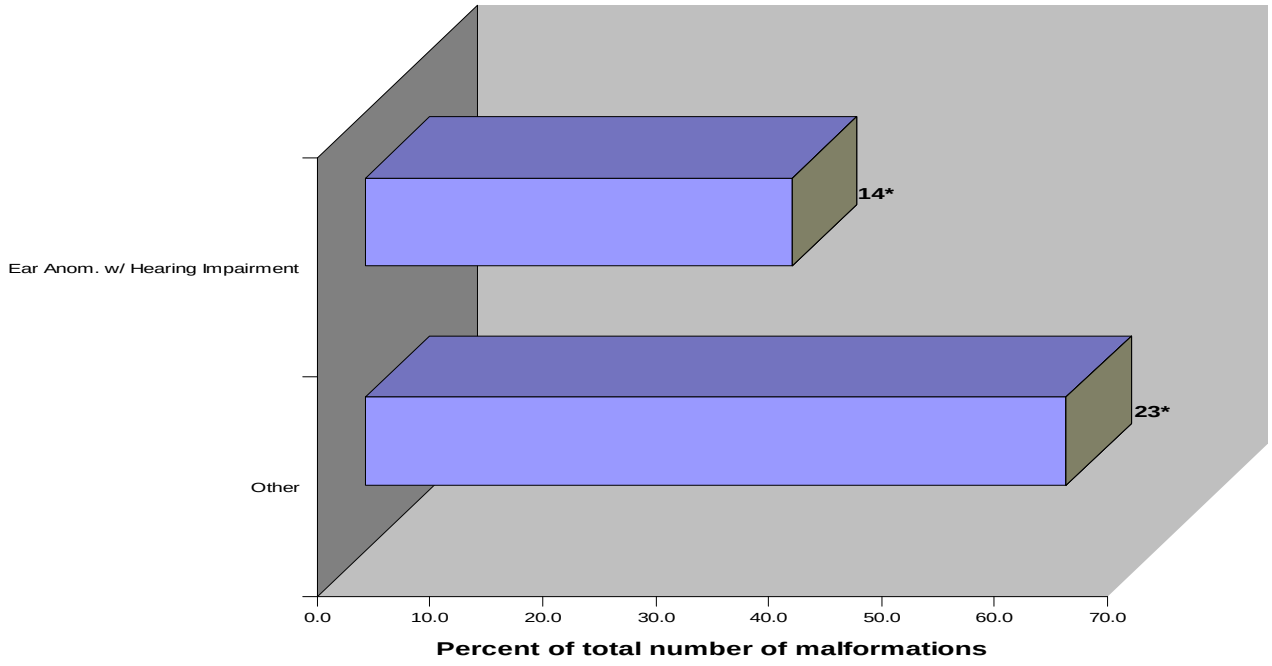
Major Malformations by Organ System
2005 Births - New York State Residents
Chromosomal Subset Category
(Number of Children = 393)
(* - Number of malformations in each organ system)



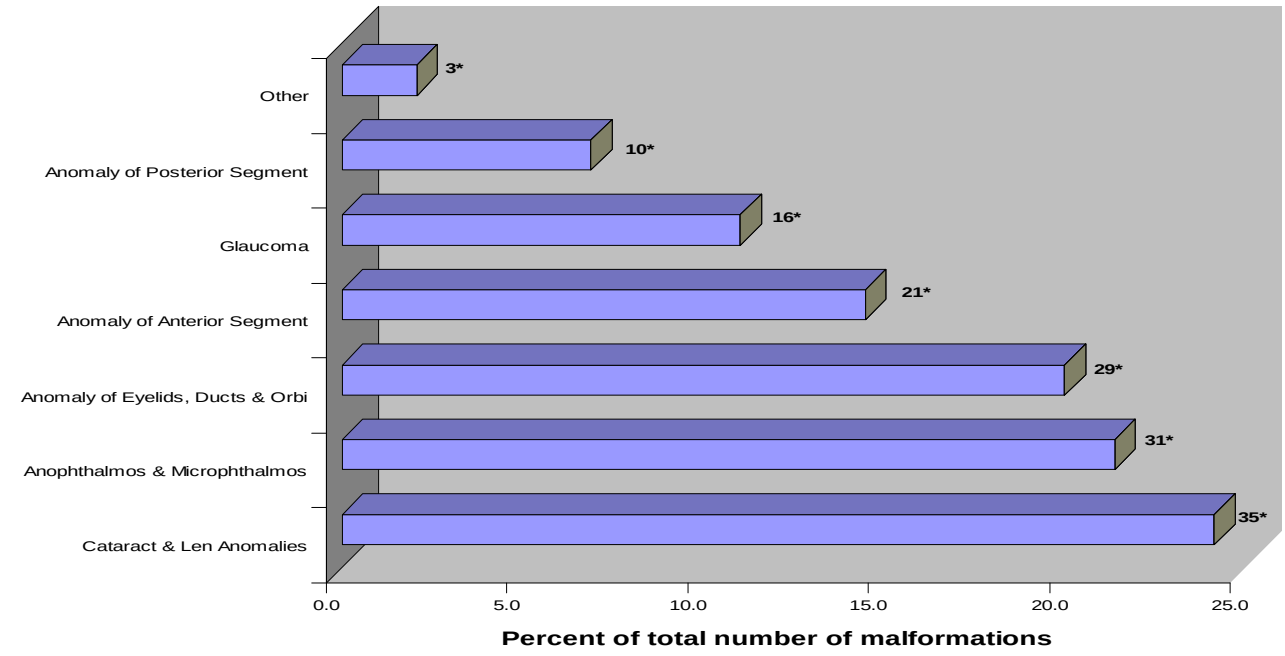

Major Malformations by Organ System
2005 Births - New York State Residents
Oral Clefts Subset Category
(Number of Children = 314)
(* - Number of malformations in each organ system)
Cleft Lip
Cleft Palate & Lip
Cleft Palate
54*
124*
136*
0.0
5.0
10.0
15.0
20.0
25.0
30.0
35.0
40.0
45.0
Percent of total number of malformations


Major Malformations by Organ System
2005 Births - New York State Residents
Digestive System Subset Category
(Number of Children = 989)
(* - Number of malformations in each organ system)
Tongue, Mouth & Pharynx Anom.
Other
Gallbladder & Liver Anom.
Tracheosophageal Fistula
Hirshsprung's Disease
A & S of Small Intestine
A & S of Large Intestine
Other Anomalies of Intestine
Pyloric Stenosis
8*
12*
52*
56*
73*
79*
108*
115*
526*
0.0
10.0
20.0
30.0
40.0
50.0
60.0
Percent of total number of malformations

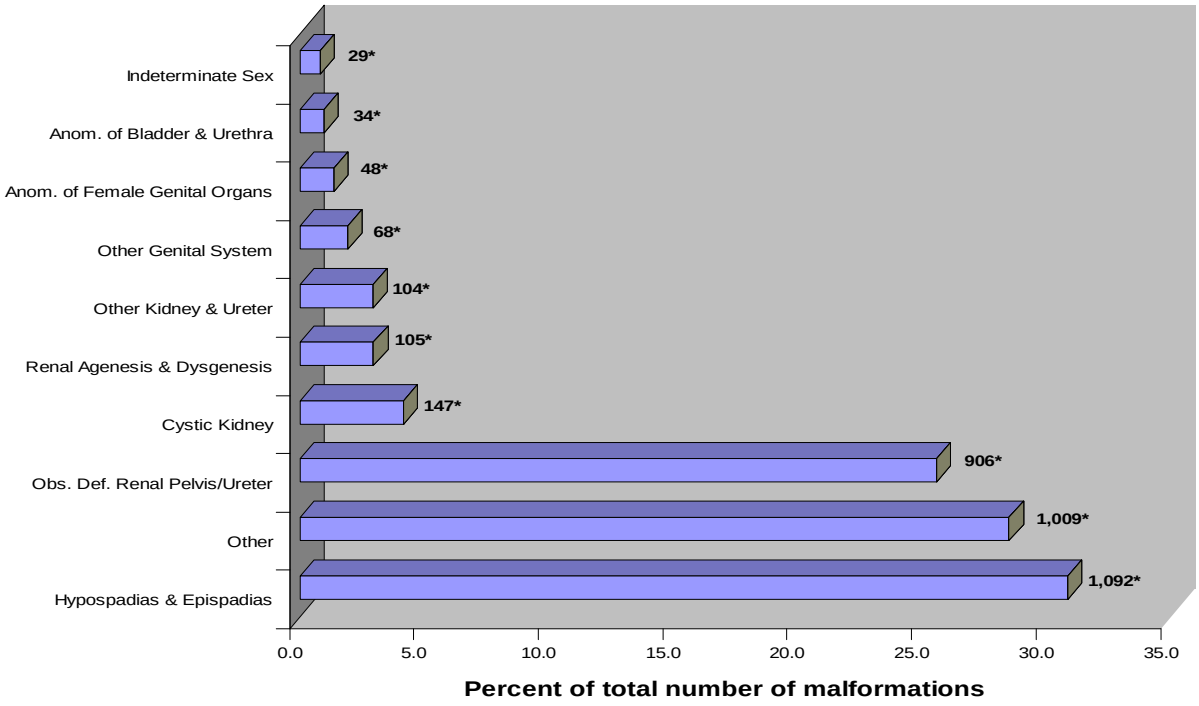
Major Malformations by Organ System
2005 Births - New York State Residents
Ear Subset Category
(Number of Children = 36)
(* - Number of malformations in each organ system)



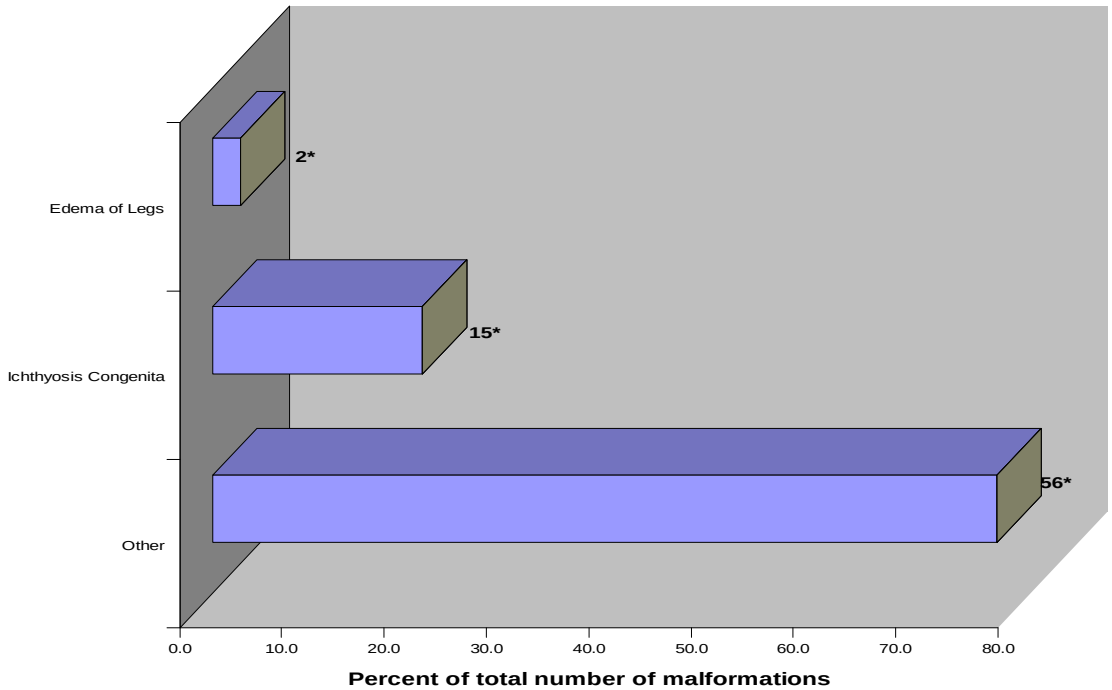
Major Malformations by Organ System
2005 Births - New York State Residents
Eye Subset Category
(Number of Children = 139)
(* - Number of malformations in each organ system)



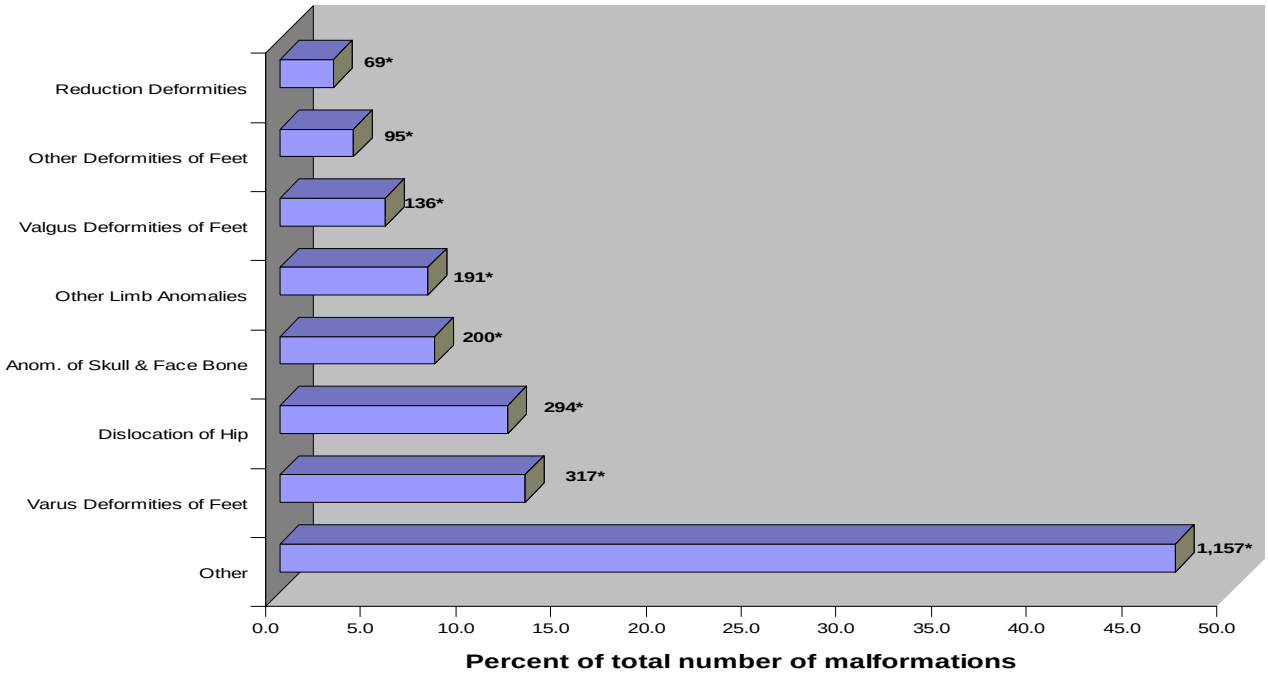
Major Malformations by Organ System
2005 Births - New York State Residents
Genitourinary System Subset Category
(Number of Children = 3,313)
(* - Number of malformations in each organ system)



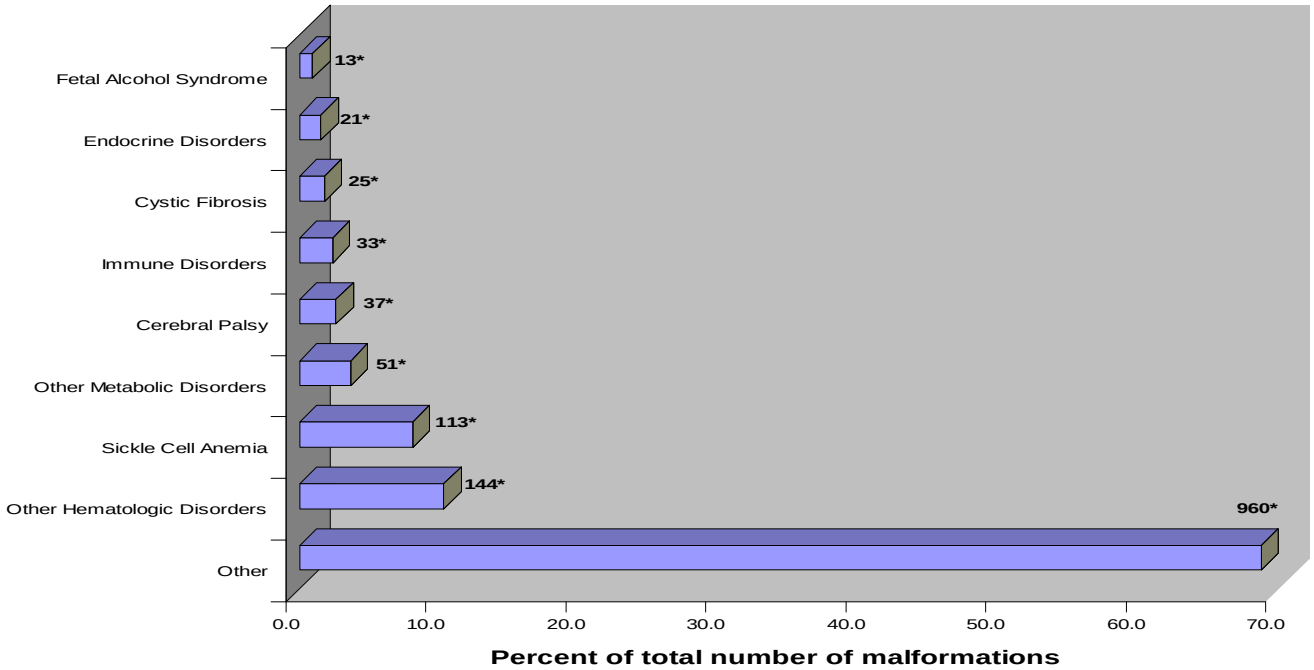
Major Malformations by Organ System
2005 Births - New York State Residents
Integument System Subset Category
(Number of Children = 73)
(* - Number of malformations in each organ system)



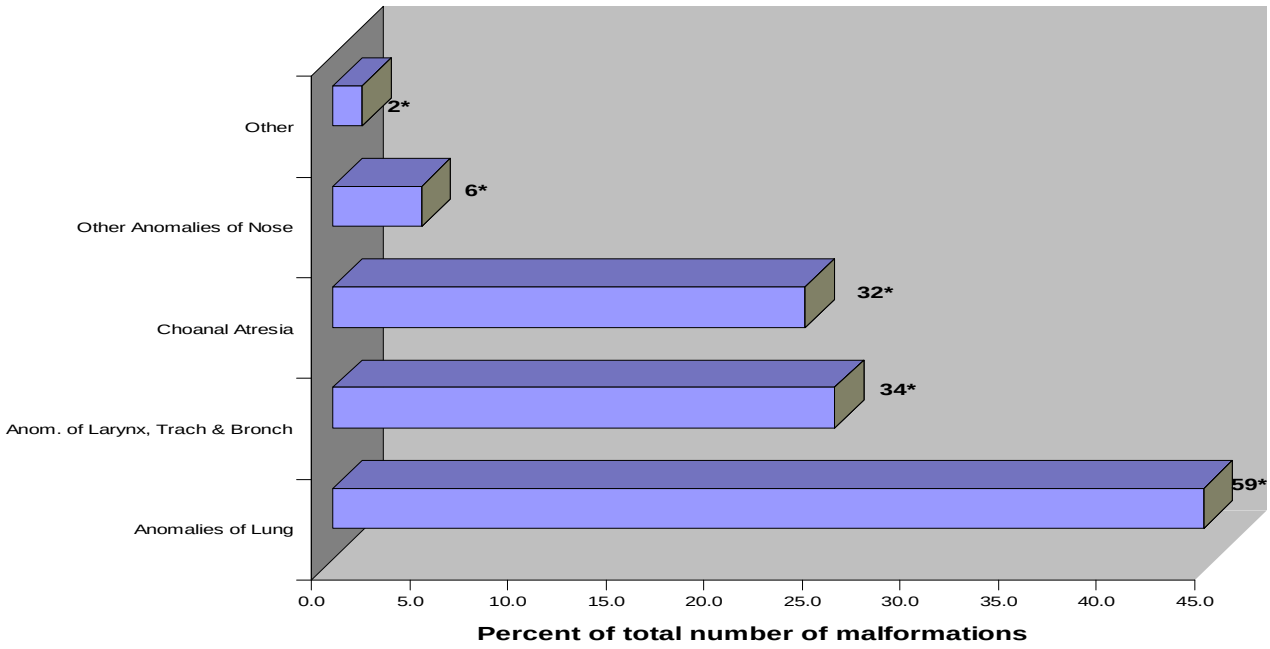
Major Malformations by Organ System
2005 Births - New York State Residents
Musculoskeletal System Subset Category
(Number of Children =2,416)
(* - Number of malformations in each organ system)



Major Malformations by Organ System
2005 Births - New York State Residents
All Others Subset Category
(Number of Children = 1,397)
(* - Number of malformations in each organ system)



Major Malformations by Organ System
2005 Births - New York State Residents
Respiratory System Subset Category
(Number of Children = 133)
(* - Number of malformations in each organ system)



Section III
Prevalence of Selected Malformations By Sex and Race/Ethnicity

Introduction to Tables

The malformations presented in this section were selected because of the frequency with which they were reported and/or their clinical significance. Rates are per 10,000 live births. The sex ratio is calculated by dividing the rate in males by the rate in females. The malformation rates presented in this report may not be comparable to earlier reports. Previous reports from 1989 to 1991 did not use birth certificate matched cases; thus, the race and birthweight from the birth certificate were not available. Birthweight data are useful to calculate the rate of some malformations such as patent ductus arteriosus. In some cases, these conditions can result from being preterm rather than actually having a malformation. Racial data in this report also may not be comparable because race is defined by maternal race from the birth certificate. In the earlier reports, race was defined by what was reported on the CMR form, which may differ from what is recorded on the birth certificate.

Fluctuations in specific malformation prevalence should be interpreted with caution, especially differences in the "other" race category since the numbers in this group are small. In addition, several malformations were added in 1992 as a result of the change to the BPA coding system. Previously, these could not be distinguished using the ICD-9 codes. However, since ICD-9 codes are more familiar to most vendors, the ICD-9 code is given on the table with the named malformation. See Appendix 4 for further information on the BPA codes.

Section III
Children With Selected Malformations
Prevalence per 10,000 Live Births by Sex and Race/Ethnicity

2005 Births– New York State Residents

ICD-9 Code	Malformation	Total Number	Total Prevalence	Male	Female	Ratio (M/F)	Non-Hispanic White	Non-Hispanic Black	His-panic	Other/ Unknown Race
243	Congenital hypothyroidism	81	3.3	3.5	3.2	1.1	3.1	5.4	2.5	2.9
270.1	Phenylketonuria	6	0.2	0.4	0.1	4.8	0.3	0.0	0.2	0.4
277.0	Cystic fibrosis	25	1.0	0.8	1.3	0.6	1.3	1.5	0.5	0.0
282.6	Sickle-cell anemia	113	4.7	4.5	4.8	0.9	0.2	23.5	2.1	0.8
658.8	Amniotic bands	1	0.0	0.1	0.0	0.0	0.0	0.0	0.2	0.0
740.0	Anencephalus	8	0.3	0.4	0.3	1.6	0.2	0.2	0.5	0.4
741.0	Spina bifida with hydrocephalus	24	1.0	1.2	0.8	1.6	0.9	2.0	0.9	0.0
741.9	Spina bifida without hydrocephalus	23	0.9	0.7	1.2	0.6	0.9	1.7	0.7	0.4
742.0	Encephalocele	12	0.5	0.7	0.3	2.9	0.5	0.5	0.7	0.0
742.1	Microcephalus	142	5.9	4.6	7.2	0.6	4.3	10.5	6.5	4.2
742.2	Agyria & lissencephaly	1	0.0	0.0	0.1	0.0	0.0	0.0	0.0	0.4
742.2	Anomalies of corpus callosum	40	1.6	1.9	1.4	1.4	1.7	2.0	1.6	0.8
742.2	Holoprosencephaly	7	0.3	0.2	0.3	0.7	0.3	0.7	0.0	0.0
742.3	Congenital hydrocephalus	162	6.7	7.4	5.9	1.2	5.4	9.1	8.3	5.0
742.4	Porencephaly	8	0.3	0.5	0.2	2.9	0.4	0.2	0.4	0.0
742.5	Congenital tethered cord	40	1.6	1.7	1.6	1.1	2.0	1.2	1.6	0.8
743.0	Anophthalmos	5	0.2	0.2	0.3	0.6	0.2	0.7	0.0	0.0
743.1	Microphthalmos	26	1.1	0.8	1.4	0.6	1.2	0.7	1.2	0.8
743.2	Glaucoma	16	0.7	0.8	0.5	1.6	0.8	0.5	0.5	0.4
743.3	Absence of lens	16	0.7	1.2	0.1	14.3	0.6	1.0	0.9	0.0
743.3	Congenital cataract	17	0.7	0.7	0.7	1.1	0.3	1.2	1.1	0.8
743.45	Aniridia	2	0.1	0.1	0.1	1.0	0.1	0.0	0.2	0.0
743.46	Coloboma of iris	7	0.3	0.2	0.4	0.4	0.5	0.0	0.2	0.0
744.0	Anotia/microtia	15	0.6	0.6	0.6	1.1	0.6	0.0	1.1	0.8
745.0	Common truncus	20	0.8	1.0	0.6	1.8	0.9	0.5	0.7	1.3
745.1	Transposition of great vessels	97	4.0	4.3	3.7	1.1	4.2	3.7	4.6	2.1
745.2	Tetralogy of Fallot	109	4.5	5.3	3.6	1.5	3.8	7.4	3.7	5.0
745.3	Common ventricle	15	0.6	0.7	0.5	1.4	0.4	0.0	1.6	0.4
745.4	Ventricular septal defect	1,011	41.7	37.6	45.9	0.8	43.2	41.9	39.3	39.3
745.5	Ostium secundum type atrial septal def.	1,102	45.4	44.6	46.3	1.0	34.6	80.2	44.7	42.7
745.6	Endocardial cushion defects	73	3.0	2.3	3.8	0.6	2.6	4.2	3.2	2.5
746.0	Atresia/stenosis of pulmonary valve	230	9.5	8.9	10.1	0.9	7.8	13.2	10.1	10.0
746.1	Tricuspid atresia/stenosis/hypoplasia	40	1.6	1.8	1.5	1.2	1.2	2.0	1.8	2.9
746.2	Ebstein's anomaly	22	0.9	1.0	0.8	1.4	0.8	1.0	1.2	0.4
746.3	Congenital stenosis of aortic valve	28	1.2	1.2	1.1	1.1	1.8	0.7	0.4	0.4
746.7	Hypoplastic left heart syndrome	57	2.3	2.8	1.9	1.5	3.0	1.5	2.3	0.8
746.85	Anomalies of coronary artery	13	0.5	0.5	0.6	0.8	0.4	1.5	0.2	0.4
747.0	Patent ductus arteriosis	844	34.8	37.0	32.5	1.1	33.9	52.2	27.8	26.3
747.10	Coartation of aorta	107	4.4	4.9	3.9	1.3	5.4	4.4	3.5	1.7
747.41	Total anomalous pulmonary venus connec	19	0.8	0.8	0.8	1.1	0.8	0.7	0.9	0.4

2005 Births– New York State Residents (continued)

ICD-9 Code	Malformation	Total Number	Total Prevalence	Male	Female	Ratio (M/F)	Non-Hispanic White	Non-Hispanic Black	His-panic	Other/ Unknown Race
748.0	Choanal atresia	32	1.3	1.4	1.3	1.1	1.5	2.0	0.7	0.8
748.5	Agenesis/hypoplasia of lung	42	1.7	2.5	0.9	2.7	2.0	2.0	1.2	1.3
749.0	Cleft palate	136	5.6	4.3	7.0	0.6	6.2	4.2	5.0	6.7
749.1	Cleft lip	54	2.2	3.0	1.4	2.1	2.5	1.0	1.8	4.2
749.2	Cleft palate & lip	124	5.1	5.6	4.6	1.2	4.9	3.7	6.2	5.9
750.3	Tracheoesophageal fistula etc.	56	2.3	3.0	1.6	1.9	2.8	1.5	2.1	1.7
750.5	Congenital hypertrophic pyloric stenosis	526	21.7	35.5	7.2	4.9	23.7	9.1	30.8	11.3
751.1	Atresia and stenosis of small intestine	79	3.3	3.6	2.9	1.3	2.5	4.4	4.1	3.3
751.2	Atresia and stenosis of rectum or anus	108	4.5	4.3	4.7	0.9	4.4	5.2	5.0	2.1
751.3	Hirschsprungs disease	73	3.0	3.9	2.1	1.8	2.5	5.2	2.8	2.5
751.4	Anomalies of intestinal fixation	43	1.8	2.2	1.4	1.6	1.4	2.5	2.1	1.7
751.61	Biliary atresia	24	1.0	0.7	1.3	0.6	0.5	1.5	1.4	1.7
752.6	Epispadias	30	1.2	2.3	0.1	27.6	1.6	1.5	0.5	0.4
752.6	Hypospadias	881	36.3	70.4	0.4	166.6	46.5	28.5	24.8	25.1
753.0	Renal agenesis and dysgenesis	105	4.3	5.5	3.0	1.8	5.3	2.9	4.2	2.1
753.1	Cystic kidney disease	147	6.1	6.3	5.8	1.1	5.7	8.1	6.4	3.8
753.2	Obstructive defect renal pelvis & ureter	906	37.3	51.2	22.7	2.3	39.8	33.1	35.2	37.2
753.5	Extrophy of urinary bladder	5	0.2	0.1	0.3	0.2	0.3	0.0	0.2	0.0
753.6	Atresia & stenosis of urethra & bladder	14	0.6	1.0	0.2	5.7	0.2	0.7	1.2	0.8
754.3	Congenital dislocation of hip	202	8.3	3.3	13.6	0.2	9.5	4.4	8.7	8.4
754.51	Talipes equinovarus	221	9.1	10.8	7.4	1.5	10.0	9.6	7.8	7.1
755.2	Reduction deformities of upper limb	45	1.9	2.1	1.6	1.3	1.8	3.2	1.4	0.8
755.3	Reduction deformities of lower limb	20	0.8	1.0	0.7	1.4	0.9	0.7	0.0	2.5
755.8	Arthrogryposis multiplex congenita	9	0.4	0.0	0.8	0.0	0.3	0.2	0.5	0.4
756.0	Craniosynostosis	117	4.8	6.0	3.6	1.7	5.4	2.7	5.7	3.3
756.0	Goldenhar syndrome	12	0.5	0.6	0.4	1.3	0.4	0.2	0.7	0.8
756.4	Chonodrodystrophy	40	1.6	2.1	1.2	1.8	1.6	2.0	1.6	1.3
756.51	Osteogenesis imperfecta	15	0.6	0.8	0.4	1.9	0.7	1.0	0.5	0.0
756.6	Diaphragmatic hernia	67	2.8	3.5	1.9	1.8	2.5	2.7	3.0	3.8
756.7	Gastroschisis	57	2.3	2.3	2.4	1.0	2.4	2.9	2.7	0.4
756.7	Omphalocele	29	1.2	1.0	1.4	0.8	1.3	2.2	0.7	0.0
756.7	Prune belly	6	0.2	0.5	0.0	0.0	0.3	0.2	0.0	0.4
758.0	Down syndrome	285	11.7	10.5	13.0	0.8	12.9	11.3	11.1	7.9
758.1	Patau syndrome	27	1.1	1.2	1.0	1.2	1.0	1.5	1.1	1.3
758.2	Edwards syndrome	19	0.8	0.5	1.1	0.4	0.7	0.7	0.5	1.7
758.6	Gonadal dysgenesis	26	1.1	0.2	1.9	0.1	1.1	1.2	1.1	0.8
758.7	Klinefelter syndrome	4	0.2	0.3	0.0	0.0	0.2	0.2	0.0	0.0
759.3	Situs inversus	17	0.7	0.9	0.5	1.7	0.3	0.5	1.4	1.3
760.71	Fetal alcohol syndrome	13	0.5	0.5	0.6	0.8	0.8	0.5	0.2	0.0
771.0	Congenital rubella	1	0.0	0.0	0.1	0.0	0.0	0.2	0.0	0.0
771.1	Congenital cytomegalovirus infection	16	0.7	0.7	0.6	1.2	0.3	1.5	0.5	1.3
771.2	Other congenital infections	61	2.5	2.7	2.4	1.1	2.5	2.2	3.5	0.8

Section IV
Most Frequently Reported Selected Major Malformations by County

Introduction to Tables

Congenital Malformations Registry data were tabulated by county of residence at the time of birth and four digit ICD-9-CM codes for major malformations. Certain codes for rare disorders and nonspecific codes are not included. The table on the next page presents the number of children with major malformations by county, and the percent of live births for comparison.

For each county, the 10 most frequently reported codes are listed, except those instances in which the tenth and subsequent codes were equal in number. In this circumstance, the additional codes of equal number are listed. Some counties may have fewer than 10 codes reported. Children reported with more than one malformation may be represented more than once in these tables. These are presented on the following pages.

These county listings are not designed to be used for comparison among counties or for analytical studies. They are most useful to assist in county planning, education, counseling and other health care services programs.

Section IV – Table 1
Children with Major Congenital Malformations & Percent of Live Births by County and Birth Year, 2005

County	Number of Children	Number of Live Births	Percent of Live Births
Albany	136	3,225	4.2
Allegany	24	501	4.8
Bronx	1,193	22,017	5.4
Broome	90	2,037	4.4
Cattaraugus	44	936	4.7
Cayuga	39	810	4.8
Chautauqua	89	1,322	6.7
Chemung	46	984	4.7
Chenango	27	548	4.9
Clinton	24	755	3.2
Columbia	18	559	3.2
Cortland	16	571	2.8
Delaware	21	443	4.7
Dutchess	129	3,002	4.3
Erie	545	9,826	5.5
Essex	13	321	4.0
Franklin	12	468	2.6
Fulton	33	539	6.1
Genesee	34	685	5.0
Greene	29	453	6.4
Herkimer	38	666	5.7
Jefferson	148	1,795	8.2
Kings	2,086	39,295	5.3
Lewis	27	369	7.3
Livingston	25	628	4.0
Madison	30	725	4.1
Monroe	340	8,473	4.0
Montgomery	37	609	6.1
Nassau	697	15,083	4.6
New York	874	20,115	4.3
Niagara	116	2,225	5.2
Oneida	146	2,479	5.9
Onondaga	292	5,346	5.5
Ontario	35	1,099	3.2
Orange	225	5,025	4.5
Orleans	17	419	4.1
Oswego	81	1,352	6.0
Otsego	21	528	4.0
Putnam	50	959	5.2
Queens	1,227	29,980	4.1
Rensselear	68	1,651	4.1
Richmond	220	5,593	3.9

Section IV – Table 1 (continued)
Children with Major Congenital Malformations & Percent of Live Births
by County and Birth Year, 2005

County	Number of Children	Number of Live Births	Percent of Live Births
Rockland	205	4,356	4.7
Saratoga	90	2,297	3.9
Schenectady	99	1,827	5.4
Schoharie	17	273	6.2
Schuyler	3	145	2.1
Seneca	13	374	3.5
St Lawrence	59	1,215	4.9
Steuben	44	1,088	4.0
Suffolk	927	18,759	4.9
Sullivan	30	845	3.6
Tioga	19	331	5.7
Tompkins	27	862	3.1
Ulster	83	1,769	4.7
Warren	32	676	4.7
Washington	33	542	6.1
Wayne	44	1,056	4.2
Westchester	545	11,076	4.9
Wyoming	35	418	8.4
Yates	9	301	3.0

Section IV - Table 2
Most Frequently Reported Major Malformations By County

Birth Year: 2005

County	ICD-9 Code	Description	Number
ALBANY	753.2	Obstructive defects of renal pelvis & ureter	18
	752.6	Hypospadias & epispadias	16
	752.5	Undescended testicle	15
	745.4	Ventricular septal defect	13
	745.5	Ostium secundum atrial septal defect	13
	747.0	Patent ductus arteriosus	10
	754.3	Congenital dislocation of hip	8
	754.6	Valgus deformities of feet	6
	755.0	Polydactyly	5
	746.6	Congenital mitral insufficiency	4
	747.1	Coarctation of aorta	4
	750.5	Congenital hypertrophic pyloric stenosis	4
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	4
ALLEGANY	752.5	Undescended testicle	4
	753.2	Obstructive defects of renal pelvis & ureter	4
	745.4	Ventricular septal defect	3
	746.8	Other specified anomalies of heart	3
	745.5	Ostium secundum atrial septal defect	2
	754.3	Congenital dislocation of hip	2
	745.1	Transposition of great vessels	1
	746.3	Congenital stenosis of aortic valve	1
	746.4	Congenital insufficiency of aortic valve	1
	747.0	Patent ductus arteriosus	1
	747.1	Coarctation of aorta	1
	749.0	Cleft palate	1
	749.1	Cleft lip	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	752.8	Other specified anomalies of genital organs	1
	753.6	Atresia and stenosis of urethra & bladder neck	1
	754.7	Other deformities of feet	1
	755.2	Reduction deformities of upper limb	1
	756.0	Anomalies of skull and face bones	1
BRONX	755.0	Polydactyly	137
	752.5	Undescended testicle	98
	745.5	Ostium secundum atrial septal defect	95
	752.6	Hypospadias & epispadias	85
	745.4	Ventricular septal defect	73
	753.2	Obstructive defects of renal pelvis & ureter	73
	747.0	Patent ductus arteriosus	66
	750.5	Congenital hypertrophic pyloric stenosis	48
	754.5	Varus deformities of feet	38
	754.3	Congenital dislocation of hip	33

Section IV - Table 2
Most Frequently Reported Major Malformations By County

Birth Year: 2005

County	ICD-9 Code	Description	Number
BROOME	745.5	Ostium secundum atrial septal defect	14
	745.4	Ventricular septal defect	8
	747.3	Anomalies of pulmonary artery	8
	755.0	Polydactyly	6
	747.0	Patent ductus arteriosus	5
	752.5	Undescended testicle	4
	752.6	Hypospadias & epispadias	4
	753.1	Cystic kidney disease	4
	750.5	Congenital hypertrophic pyloric stenosis	3
	754.5	Varus deformities of feet	3
CATTARAUGUS	746.8	Other specified anomalies of heart	4
	333.2	Myoclonus	3
	745.4	Ventricular septal defect	3
	745.5	Ostium secundum atrial septal defect	3
	747.0	Patent ductus arteriosus	3
	752.5	Undescended testicle	3
	752.6	Hypospadias & epispadias	3
	756.6	Anomalies of diaphragm	3
	756.7	Anomalies of abdominal wall	3
	750.5	Congenital hypertrophic pyloric stenosis	2
	755.0	Polydactyly	2
	756.0	Anomalies of skull and face bones	2
CAYUGA	745.4	Ventricular septal defect	5
	746.0	Anomalies of pulmonary valve	4
	747.0	Patent ductus arteriosus	4
	752.6	Hypospadias & epispadias	4
	755.6	Other anomalies of lower limb including pelvic girdle	4
	752.5	Undescended testicle	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.3	Congenital dislocation of hip	2
	755.0	Polydactyly	2
	343.9	Infantile cerebral palsy unspecified	1
	743.6	Congenital anomalies of eyelids, lacrimal system & orbit	1
	745.5	Ostium secundum atrial septal defect	1
	746.8	Other specified anomalies of heart	1
	747.1	Coarctation of aorta	1
	747.3	Anomalies of pulmonary artery	1
	747.8	Other specified anomalies of circulatory system	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.5	Other anomalies of intestine	1
	753.1	Cystic kidney disease	1
	754.5	Varus deformities of feet	1
	754.6	Valgus deformities of feet	1
	756.0	Anomalies of skull and face bones	1
	758.0	Down syndrome	1

Section IV - Table 2
Most Frequently Reported Major Malformations By County

Birth Year: 2005

County	ICD-9 Code	Description	Number
CHAUTAUQUA	745.5	Ostium secundum atrial septal defect	23
	745.4	Ventricular septal defect	15
	746.8	Other specified anomalies of heart	7
	752.5	Undescended testicle	7
	747.0	Patent ductus arteriosus	6
	747.3	Anomalies of pulmonary artery	6
	749.0	Cleft palate	4
	750.5	Congenital hypertrophic pyloric stenosis	4
	752.6	Hypospadias & epispadias	4
	753.2	Obstructive defects of renal pelvis & ureter	4
	756.0	Anomalies of skull and face bones	4
CHEMUNG	753.2	Obstructive defects of renal pelvis & ureter	8
	752.6	Hypospadias & epispadias	6
	750.5	Congenital hypertrophic pyloric stenosis	4
	752.5	Undescended testicle	3
	273.8	Other disorders of plasma protien	2
	745.4	Ventricular septal defect	2
	745.5	Ostium secundum atrial septal defect	2
	749.2	Cleft palate with cleft lip	2
	228.0	Hemangioma, any site	1
	228.1	Lymphangioma, any site	1
	271.1	Galactosemia	1
	740.0	Anencephalus	1
	742.3	Congenital hydrocephalus	1
	745.2	Tetralogy of Fallot	1
	747.0	Patent ductus arteriosus	1
	747.3	Anomalies of pulmonary artery	1
	748.0	Choanal atresia	1
	749.0	Cleft palate	1
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	751.5	Other anomalies of intestine	1
	751.6	Anomalies of gallbladder, bile ducts, and liver	1
	753.0	Renal agenesis & dysgenesis	1
	753.1	Cystic kidney disease	1
	754.3	Congenital dislocation of hip	1
	754.7	Other deformities of feet	1
	755.0	Polydactyly	1
	756.4	Chondrodystrophy	1
	771.2	Other congenital infections	1
CHENANGO	750.5	Congenital hypertrophic pyloric stenosis	4
	747.0	Patent ductus arteriosus	3
	752.5	Undescended testicle	3
	752.6	Hypospadias & epispadias	3
	754.5	Varus deformities of feet	3

Section IV - Table 2
Most Frequently Reported Major Malformations By County

Birth Year: 2005

County	ICD-9 Code	Description	Number
CHENANGO	754.3	Congenital dislocation of hip	2
	524.0	Major anomalies of jaw size	1
	741.0	Spina bifida with hydrocephalus	1
	741.9	Spina bifida w/o hydrocephalus	1
	742.3	Congenital hydrocephalus	1
	742.5	Other specified anomalies of spinal cord	1
	745.4	Ventricular septal defect	1
	745.5	Ostium secundum atrial septal defect	1
	745.6	Endocardial cushion defects	1
	746.4	Congenital insufficiency of aortic valve	1
	753.7	Anomalies of urachus	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
	756.0	Anomalies of skull and face bones	1
	756.1	Anomalies of spine	1
	756.7	Anomalies of abdominal wall	1
	758.2	Edwards syndrome	1
CLINTON	745.4	Ventricular septal defect	4
	745.5	Ostium secundum atrial septal defect	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	745.2	Tetralogy of Fallot	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	752.5	Undescended testicle	2
	752.6	Hypospadias & epispadias	2
	752.8	Other specified anomalies of genital organs	2
	343.9	Infantile cerebral palsy unspecified	1
	744.0	Anomalies of ear causing impairment of hearing	1
	746.0	Anomalies of pulmonary valve	1
	746.1	Tricuspid atresia & stenosis	1
	746.8	Other specified anomalies of heart	1
	747.0	Patent ductus arteriosus	1
	749.0	Cleft palate	1
	749.2	Cleft palate with cleft lip	1
	755.2	Reduction deformities of upper limb	1
	757.3	Other specified anomalies of skin	1
COLUMBIA	753.2	Obstructive defects of renal pelvis & ureter	4
	752.6	Hypospadias & epispadias	3
	755.0	Polydactyly	3
	750.5	Congenital hypertrophic pyloric stenosis	2
	255.2	Adrenogenital disorders	1
	745.4	Ventricular septal defect	1
	745.5	Ostium secundum atrial septal defect	1
	747.0	Patent ductus arteriosus	1
	747.3	Anomalies of pulmonary artery	1
	748.5	Agenesis, hypoplasia & dysplasia, lung	1

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Most Frequently Reported Major Malformations By County

Birth Year: 2005

County	ICD-9 Code	Description	Number
COLUMBIA	749.2	Cleft palate with cleft lip	1
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	1
	751.5	Other anomalies of intestine	1
	754.3	Congenital dislocation of hip	1
	756.1	Anomalies of spine	1
	759.8	Other specified anomalies	1
CORTLAND	752.5	Undescended testicle	3
	752.6	Hypospadias & epispadias	3
	758.0	Down syndrome	2
	745.5	Ostium secundum atrial septal defect	1
	746.4	Congenital insufficiency of aortic valve	1
	746.8	Other specified anomalies of heart	1
	747.0	Patent ductus arteriosus	1
	749.0	Cleft palate	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.5	Varus deformities of feet	1
	755.0	Polydactyly	1
	756.1	Anomalies of spine	1
	759.8	Other specified anomalies	1
DELAWARE	752.5	Undescended testicle	3
	750.5	Congenital hypertrophic pyloric stenosis	2
	743.1	Microphthalmos	1
	744.2	Other specified anomalies of ear	1
	745.3	Common ventricle	1
	745.5	Ostium secundum atrial septal defect	1
	746.4	Congenital insufficiency of aortic valve	1
	746.7	Hypoplastic left heart syndrome	1
	746.8	Other specified anomalies of heart	1
	747.0	Patent ductus arteriosus	1
	747.4	Anomalies of great veins	1
	748.1	Other anomalies of nose	1
	749.1	Cleft lip	1
	751.1	Atresia & stenosis of small intestine	1
	752.4	Anomalies of cervix, vagina & external female genitalia	1
	752.6	Hypospadias & epispadias	1
	753.4	Other specified anomalies of ureter	1
	754.4	Congenital genu recurvatum & bowing of long bones of leg	1
	754.7	Other deformities of feet	1
	755.0	Polydactyly	1
	755.2	Reduction deformities of upper limb	1
	756.7	Anomalies of abdominal wall	1
	757.1	Ichthyosis congenita	1

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Birth Year: 2005

County	ICD-9 Code	Description	Number
DELAWARE	759.0	Anomalies of spleen	1
	759.3	Situs inversus	1
	771.2	Other congenital infections	1
DUTCHESS	752.5	Undescended testicle	16
	745.4	Ventricular septal defect	13
	752.6	Hypospadias & epispadias	12
	750.5	Congenital hypertrophic pyloric stenosis	9
	747.0	Patent ductus arteriosus	7
	754.3	Congenital dislocation of hip	7
	758.0	Down syndrome	7
	753.2	Obstructive defects of renal pelvis & ureter	6
	746.8	Other specified anomalies of heart	4
	751.4	Anomalies of intestinal fixation	4
ERIE	747.0	Patent ductus arteriosus	70
	752.5	Undescended testicle	52
	745.4	Ventricular septal defect	46
	752.6	Hypospadias & epispadias	40
	746.8	Other specified anomalies of heart	35
	755.0	Polydactyly	32
	747.3	Anomalies of pulmonary artery	29
	753.2	Obstructive defects of renal pelvis & ureter	27
	750.5	Congenital hypertrophic pyloric stenosis	25
	745.5	Ostium secundum atrial septal defect	22
ESSEX	747.0	Patent ductus arteriosus	3
	745.4	Ventricular septal defect	2
	752.5	Undescended testicle	2
	742.2	Reduction deformities of brain	1
	742.5	Other specified anomalies of spinal cord	1
	743.3	Congenital cataract & lens anomalies	1
	745.5	Ostium secundum atrial septal defect	1
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	1
	752.4	Anomalies of cervix, vagina & external female genitalia	1
	752.6	Hypospadias & epispadias	1
	753.0	Renal agenesis & dysgenesis	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	756.3	Other anomalies of ribs and sternum	1
	759.8	Other specified anomalies	1
FRANKLIN	752.6	Hypospadias & epispadias	4
	752.5	Undescended testicle	3
	745.4	Ventricular septal defect	1
	746.0	Anomalies of pulmonary valve	1
	746.8	Other specified anomalies of heart	1

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Birth Year: 2005

County	ICD-9 Code	Description	Number
FRANKLIN	747.0	Patent ductus arteriosus	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	756.0	Anomalies of skull and face bones	1
FULTON	752.5	Undescended testicle	8
	752.6	Hypospadias & epispadias	4
	754.3	Congenital dislocation of hip	4
	754.6	Valgus deformities of feet	3
	742.3	Congenital hydrocephalus	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	742.1	Microcephalus	1
	745.1	Transposition of great vessels	1
	745.3	Common ventricle	1
	745.5	Ostium secundum atrial septal defect	1
	747.0	Patent ductus arteriosus	1
	747.1	Coarctation of aorta	1
	747.3	Anomalies of pulmonary artery	1
	747.6	Other anomalies of peripheral vascular system	1
	751.3	Hirschprung's disease & other functional disorders of colon	1
	752.7	Indeterminate sex & pseudo-hermaphroditism	1
	755.0	Polydactyly	1
	755.1	Syndactyly	1
	756.7	Anomalies of abdominal wall	1
GENESEE	752.6	Hypospadias & epispadias	7
	745.4	Ventricular septal defect	3
	747.0	Patent ductus arteriosus	3
	745.5	Ostium secundum atrial septal defect	2
	746.8	Other specified anomalies of heart	2
	752.5	Undescended testicle	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.1	Deformities of sternocleidomastoid muscle	2
	755.0	Polydactyly	2
	756.0	Anomalies of skull and face bones	2
GREENE	755.0	Polydactyly	5
	752.6	Hypospadias & epispadias	3
	745.4	Ventricular septal defect	2
	749.2	Cleft palate with cleft lip	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	752.5	Undescended testicle	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.5	Varus deformities of feet	2
	758.0	Down syndrome	2
	255.2	Adrenogenital disorders	1

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Most Frequently Reported Major Malformations By County

Birth Year: 2005

County	ICD-9 Code	Description	Number
GREENE	282.6	Sickle-cell anemia	1
	742.4	Other specified anomalies of brain	1
	745.5	Ostium secundum atrial septal defect	1
	747.0	Patent ductus arteriosus	1
	747.2	Other anomalies of aorta	1
	751.1	Atresia & stenosis of small intestine	1
	751.5	Other anomalies of intestine	1
	753.1	Cystic kidney disease	1
	756.4	Chondrodystrophy	1
	756.7	Anomalies of abdominal wall	1
HERKIMER	752.6	Hypospadias & epispadias	7
	742.3	Congenital hydrocephalus	2
	745.5	Ostium secundum atrial septal defect	2
	747.3	Anomalies of pulmonary artery	2
	752.5	Undescended testicle	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.5	Varus deformities of feet	2
	754.6	Valgus deformities of feet	2
	755.6	Other anomalies of lower limb including pelvic girdle	2
	756.7	Anomalies of abdominal wall	2
JEFFERSON	754.5	Varus deformities of feet	26
	752.6	Hypospadias & epispadias	14
	745.4	Ventricular septal defect	13
	752.5	Undescended testicle	13
	756.0	Anomalies of skull and face bones	6
	750.5	Congenital hypertrophic pyloric stenosis	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	745.5	Ostium secundum atrial septal defect	4
	756.1	Anomalies of spine	4
KINGS	745.5	Ostium secundum atrial septal defect	328
	747.0	Patent ductus arteriosus	235
	745.4	Ventricular septal defect	201
	752.5	Undescended testicle	162
	755.0	Polydactyly	156
	752.6	Hypospadias & epispadias	137
	753.2	Obstructive defects of renal pelvis & ureter	135
	746.8	Other specified anomalies of heart	90
	750.5	Congenital hypertrophic pyloric stenosis	75
	747.3	Anomalies of pulmonary artery	51
LEWIS	752.6	Hypospadias & epispadias	3
	524.0	Major anomalies of jaw size	2
	746.8	Other specified anomalies of heart	2

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Most Frequently Reported Major Malformations By County

Birth Year: 2005

County	ICD-9 Code	Description	Number
LEWIS	749.0	Cleft palate	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.5	Varus deformities of feet	2
	758.0	Down syndrome	2
	742.1	Microcephalus	1
	745.1	Transposition of great vessels	1
	745.2	Tetralogy of Fallot	1
	745.5	Ostium secundum atrial septal defect	1
	745.6	Endocardial cushion defects	1
	746.6	Congenital mitral insufficiency	1
	747.0	Patent ductus arteriosus	1
	747.3	Anomalies of pulmonary artery	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	753.0	Renal agenesis & dysgenesis	1
	754.7	Other deformities of feet	1
	755.0	Polydactyly	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
	756.0	Anomalies of skull and face bones	1
	756.1	Anomalies of spine	1
	759.6	Other hamartoses,nec	1
LIVINGSTON	747.0	Patent ductus arteriosus	5
	752.5	Undescended testicle	3
	758.0	Down syndrome	3
	750.5	Congenital hypertrophic pyloric stenosis	2
	752.6	Hypospadias & epispadias	2
	755.6	Other anomalies of lower limb including pelvic girdle	2
	756.0	Anomalies of skull and face bones	2
	277.0	Cystic fibrosis	1
	745.5	Ostium secundum atrial septal defect	1
	745.6	Endocardial cushion defects	1
	746.0	Anomalies of pulmonary valve	1
	746.1	Tricuspid atresia & stenosis	1
	746.6	Congenital mitral insufficiency	1
	746.8	Other specified anomalies of heart	1
	747.1	Coarctation of aorta	1
	751.1	Atresia & stenosis of small intestine	1
	754.3	Congenital dislocation of hip	1
	756.7	Anomalies of abdominal wall	1
MADISON	752.5	Undescended testicle	4
	745.4	Ventricular septal defect	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	752.6	Hypospadias & epispadias	2
	243.	Congenital hypothyroidism	1
	742.0	Encephalocele	1

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Birth Year: 2005

County	ICD-9 Code	Description	Number
MADISON	742.1	Microcephalus	1
	742.2	Reduction deformities of brain	1
	742.3	Congenital hydrocephalus	1
	745.2	Tetralogy of Fallot	1
	745.5	Ostium secundum atrial septal defect	1
	746.0	Anomalies of pulmonary valve	1
	746.1	Tricuspid atresia & stenosis	1
	746.3	Congenital stenosis of aortic valve	1
	746.7	Hypoplastic left heart syndrome	1
	746.8	Other specified anomalies of heart	1
	747.0	Patent ductus arteriosus	1
	747.1	Coarctation of aorta	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.1	Deformities of sternocleidomastoid muscle	1
	754.3	Congenital dislocation of hip	1
	754.5	Varus deformities of feet	1
	754.6	Valgus deformities of feet	1
	755.0	Polydactyly	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
	756.0	Anomalies of skull and face bones	1
	756.1	Anomalies of spine	1
	756.7	Anomalies of abdominal wall	1
	758.0	Down syndrome	1
	771.2	Other congenital infections	1
MONROE	752.6	Hypospadias & epispadias	52
	753.2	Obstructive defects of renal pelvis & ureter	44
	752.5	Undescended testicle	31
	745.4	Ventricular septal defect	29
	755.0	Polydactyly	18
	747.0	Patent ductus arteriosus	16
	745.5	Ostium secundum atrial septal defect	15
	750.5	Congenital hypertrophic pyloric stenosis	15
	758.0	Down syndrome	12
	754.5	Varus deformities of feet	11
MONTGOMERY	752.5	Undescended testicle	4
	750.5	Congenital hypertrophic pyloric stenosis	3
	752.6	Hypospadias & epispadias	3
	754.5	Varus deformities of feet	3
	228.0	Hemangioma, any site	2
	746.4	Congenital insufficiency of aortic valve	2
	746.7	Hypoplastic left heart syndrome	2
	747.0	Patent ductus arteriosus	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.3	Congenital dislocation of hip	2
	758.0	Down syndrome	2

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Birth Year: 2005

County	ICD-9 Code	Description	Number
NASSAU	752.6	Hypospadias & epispadias	109
	753.2	Obstructive defects of renal pelvis & ureter	85
	747.0	Patent ductus arteriosus	78
	745.4	Ventricular septal defect	55
	752.5	Undescended testicle	46
	745.5	Ostium secundum atrial septal defect	40
	750.5	Congenital hypertrophic pyloric stenosis	36
	746.8	Other specified anomalies of heart	26
	755.0	Polydactyly	21
	758.0	Down syndrome	21
NEW YORK	745.5	Ostium secundum atrial septal defect	115
	745.4	Ventricular septal defect	109
	752.5	Undescended testicle	67
	753.2	Obstructive defects of renal pelvis & ureter	60
	752.6	Hypospadias & epispadias	59
	755.0	Polydactyly	50
	747.0	Patent ductus arteriosus	44
	754.3	Congenital dislocation of hip	32
	747.3	Anomalies of pulmonary artery	29
	750.5	Congenital hypertrophic pyloric stenosis	26
NIAGARA	745.4	Ventricular septal defect	9
	745.5	Ostium secundum atrial septal defect	9
	747.0	Patent ductus arteriosus	9
	746.0	Anomalies of pulmonary valve	8
	752.5	Undescended testicle	8
	753.2	Obstructive defects of renal pelvis & ureter	8
	746.8	Other specified anomalies of heart	7
	750.5	Congenital hypertrophic pyloric stenosis	7
	755.0	Polydactyly	7
	756.0	Anomalies of skull and face bones	6
ONEIDA	752.6	Hypospadias & epispadias	19
	745.4	Ventricular septal defect	12
	752.5	Undescended testicle	10
	747.0	Patent ductus arteriosus	9
	755.6	Other anomalies of lower limb including pelvic girdle	8
	754.3	Congenital dislocation of hip	7
	755.0	Polydactyly	7
	750.5	Congenital hypertrophic pyloric stenosis	6
	756.0	Anomalies of skull and face bones	6
	753.0	Renal agenesis & dysgenesis	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	754.5	Varus deformities of feet	5

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Birth Year: 2005

County	ICD-9 Code	Description	Number
ONONDAGA	752.5	Undescended testicle	37
	752.6	Hypospadias & epispadias	36
	745.4	Ventricular septal defect	20
	753.2	Obstructive defects of renal pelvis & ureter	20
	756.1	Anomalies of spine	15
	745.5	Ostium secundum atrial septal defect	13
	755.0	Polydactyly	13
	747.0	Patent ductus arteriosus	9
	750.5	Congenital hypertrophic pyloric stenosis	9
	754.5	Varus deformities of feet	9
ONTARIO	752.6	Hypospadias & epispadias	6
	753.2	Obstructive defects of renal pelvis & ureter	6
	752.5	Undescended testicle	4
	747.0	Patent ductus arteriosus	3
	750.5	Congenital hypertrophic pyloric stenosis	3
	751.1	Atresia & stenosis of small intestine	3
	758.0	Down syndrome	3
	228.0	Hemangioma, any site	1
	282.0	Hereditary spherocytosis	1
	742.5	Other specified anomalies of spinal cord	1
	745.4	Ventricular septal defect	1
	745.5	Ostium secundum atrial septal defect	1
	745.6	Endocardial cushion defects	1
	746.0	Anomalies of pulmonary valve	1
	746.8	Other specified anomalies of heart	1
	748.5	Agenesis, hypoplasia & dysplasia, lung	1
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	1
	751.3	Hirschprung's disease & other functional disorders of colon	1
	751.4	Anomalies of intestinal fixation	1
	752.0	Anomalies of ovaries	1
	753.0	Renal agenesis & dysgenesis	1
	754.1	Deformities of sternocleidomastoid muscle	1
	755.6	Other anomalies of lower limb including pelvic girdle	1
ORANGE	745.4	Ventricular septal defect	21
	752.6	Hypospadias & epispadias	21
	753.2	Obstructive defects of renal pelvis & ureter	21
	745.5	Ostium secundum atrial septal defect	18
	752.5	Undescended testicle	16
	747.0	Patent ductus arteriosus	13
	755.0	Polydactyly	13
	750.5	Congenital hypertrophic pyloric stenosis	12
	756.0	Anomalies of skull and face bones	8
	758.0	Down syndrome	8

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Birth Year: 2005

County	ICD-9 Code	Description	Number
ORLEANS	745.5	Ostium secundum atrial septal defect	3
	747.0	Patent ductus arteriosus	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	745.4	Ventricular septal defect	2
	746.8	Other specified anomalies of heart	2
	752.5	Undescended testicle	2
	277.0	Cystic fibrosis	1
	343.9	Infantile cerebral palsy unspecified	1
	742.4	Other specified anomalies of brain	1
	747.3	Anomalies of pulmonary artery	1
	748.3	Other anomalies of larynx, trachea, & bronchus	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.6	Anomalies of gallbladder, bile ducts, and liver	1
	752.6	Hypospadias & epispadias	1
	754.7	Other deformities of feet	1
	756.7	Anomalies of abdominal wall	1
	757.6	Specified anomalies of breast	1
	771.2	Other congenital infections	1
OSWEGO	745.4	Ventricular septal defect	10
	747.0	Patent ductus arteriosus	8
	750.5	Congenital hypertrophic pyloric stenosis	6
	752.5	Undescended testicle	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	756.1	Anomalies of spine	5
	752.6	Hypospadias & epispadias	4
	746.4	Congenital insufficiency of aortic valve	3
	753.0	Renal agenesis & dysgenesis	3
	753.3	Other specified anomalies of kidney	3
	755.0	Polydactyly	3
OTSEGO	752.6	Hypospadias & epispadias	4
	745.5	Ostium secundum atrial septal defect	2
	746.4	Congenital insufficiency of aortic valve	2
	753.0	Renal agenesis & dysgenesis	2
	753.1	Cystic kidney disease	2
	286.3	Congenital deficiency of other clotting factors	1
	745.2	Tetralogy of Fallot	1
	745.4	Ventricular septal defect	1
	746.0	Anomalies of pulmonary valve	1
	746.3	Congenital stenosis of aortic valve	1
	746.6	Congenital mitral insufficiency	1
	746.8	Other specified anomalies of heart	1
	747.0	Patent ductus arteriosus	1

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Birth Year: 2005

County	ICD-9 Code	Description	Number
OTSEGO	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	752.5	Undescended testicle	1
	752.8	Other specified anomalies of genital organs	1
	756.0	Anomalies of skull and face bones	1
	759.1	Anomalies of adrenal gland	1
PUTNAM	745.5	Ostium secundum atrial septal defect	7
	752.6	Hypospadias & epispadias	5
	756.0	Anomalies of skull and face bones	5
	745.4	Ventricular septal defect	4
	750.5	Congenital hypertrophic pyloric stenosis	4
	752.5	Undescended testicle	4
	753.2	Obstructive defects of renal pelvis & ureter	4
	746.8	Other specified anomalies of heart	3
	747.0	Patent ductus arteriosus	3
	746.0	Anomalies of pulmonary valve	2
	751.3	Hirschprung's disease & other functional disorders of colon	2
	754.5	Varus deformities of feet	2
	759.8	Other specified anomalies	2
QUEENS	745.5	Ostium secundum atrial septal defect	145
	745.4	Ventricular septal defect	111
	753.2	Obstructive defects of renal pelvis & ureter	110
	752.5	Undescended testicle	101
	752.6	Hypospadias & epispadias	97
	755.0	Polydactyly	92
	747.0	Patent ductus arteriosus	60
	750.5	Congenital hypertrophic pyloric stenosis	54
	754.3	Congenital dislocation of hip	47
	758.0	Down syndrome	36
RENSSELAER	752.6	Hypospadias & epispadias	8
	753.2	Obstructive defects of renal pelvis & ureter	8
	745.4	Ventricular septal defect	4
	746.8	Other specified anomalies of heart	4
	754.3	Congenital dislocation of hip	4
	745.5	Ostium secundum atrial septal defect	3
	746.0	Anomalies of pulmonary valve	3
	747.0	Patent ductus arteriosus	3
	752.5	Undescended testicle	3
	753.4	Other specified anomalies of ureter	3
	755.0	Polydactyly	3
	756.0	Anomalies of skull and face bones	3
	758.0	Down syndrome	3

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Birth Year: 2005

County	ICD-9 Code	Description	Number
RICHMOND	752.5	Undescended testicle	26
	752.6	Hypospadias & epispadias	22
	745.4	Ventricular septal defect	16
	753.2	Obstructive defects of renal pelvis & ureter	16
	745.5	Ostium secundum atrial septal defect	13
	750.5	Congenital hypertrophic pyloric stenosis	13
	747.0	Patent ductus arteriosus	11
	755.0	Polydactyly	11
	228.0	Hemangioma, any site	7
	742.3	Congenital hydrocephalus	7
	758.0	Down syndrome	7
ROCKLAND	745.5	Ostium secundum atrial septal defect	28
	752.6	Hypospadias & epispadias	24
	752.5	Undescended testicle	23
	753.2	Obstructive defects of renal pelvis & ureter	20
	745.4	Ventricular septal defect	18
	747.0	Patent ductus arteriosus	11
	755.0	Polydactyly	10
	750.5	Congenital hypertrophic pyloric stenosis	9
	758.0	Down syndrome	9
	746.0	Anomalies of pulmonary valve	8
	754.5	Varus deformities of feet	8
SARATOGA	747.0	Patent ductus arteriosus	10
	752.5	Undescended testicle	8
	754.3	Congenital dislocation of hip	8
	752.6	Hypospadias & epispadias	7
	753.2	Obstructive defects of renal pelvis & ureter	7
	745.4	Ventricular septal defect	5
	750.5	Congenital hypertrophic pyloric stenosis	4
	753.0	Renal agenesis & dysgenesis	4
	754.5	Varus deformities of feet	4
	747.1	Coarctation of aorta	3
	758.0	Down syndrome	3
	771.2	Other congenital infections	3
SCHENECTADY	752.5	Undescended testicle	16
	752.6	Hypospadias & epispadias	14
	753.2	Obstructive defects of renal pelvis & ureter	12
	745.4	Ventricular septal defect	9
	755.0	Polydactyly	7
	754.5	Varus deformities of feet	6
	750.5	Congenital hypertrophic pyloric stenosis	5
	756.7	Anomalies of abdominal wall	4
	758.0	Down syndrome	4
	759.8	Other specified anomalies	4

Section IV - Table 2
Most Frequently Reported Major Malformations By County

Birth Year: 2005

County	ICD-9 Code	Description	Number
SCHOHARIE	752.6	Hypospadias & epispadias	3
	745.4	Ventricular septal defect	2
	752.5	Undescended testicle	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.5	Varus deformities of feet	2
	742.4	Other specified anomalies of brain	1
	745.5	Ostium secundum atrial septal defect	1
	746.0	Anomalies of pulmonary valve	1
	746.4	Congenital insufficiency of aortic valve	1
	746.8	Other specified anomalies of heart	1
	749.1	Cleft lip	1
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	753.1	Cystic kidney disease	1
	754.3	Congenital dislocation of hip	1
	756.6	Anomalies of diaphragm	1
	758.0	Down syndrome	1
SCHUYLER	746.8	Other specified anomalies of heart	1
	752.6	Hypospadias & epispadias	1
	752.8	Other specified anomalies of genital organs	1
SENECA	750.5	Congenital hypertrophic pyloric stenosis	3
	752.5	Undescended testicle	2
	273.8	Other disorders of plasma protien	1
	743.0	Anophthalmos	1
	745.4	Ventricular septal defect	1
	746.1	Tricuspid atresia & stenosis	1
	747.3	Anomalies of pulmonary artery	1
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	751.5	Other anomalies of intestine	1
	753.2	Obstructive defects of renal pelvis & ureter	1
	754.7	Other deformities of feet	1
	755.0	Polydactyly	1
	756.6	Anomalies of diaphragm	1
	758.0	Down syndrome	1
ST LAWRENCE	752.6	Hypospadias & epispadias	14
	745.4	Ventricular septal defect	5
	750.5	Congenital hypertrophic pyloric stenosis	5
	745.5	Ostium secundum atrial septal defect	3
	746.8	Other specified anomalies of heart	3
	752.5	Undescended testicle	3
	753.1	Cystic kidney disease	3

Section IV - Table 2
Most Frequently Reported Major Malformations By County

Birth Year: 2005

County	ICD-9 Code	Description	Number
ST LAWRENCE	758.0	Down syndrome	3
	745.1	Transposition of great vessels	2
	746.4	Congenital insufficiency of aortic valve	2
	749.2	Cleft palate with cleft lip	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.5	Varus deformities of feet	2
	754.6	Valgus deformities of feet	2
STEUBEN	750.5	Congenital hypertrophic pyloric stenosis	6
	752.6	Hypospadias & epispadias	6
	752.5	Undescended testicle	4
	745.4	Ventricular septal defect	3
	758.0	Down syndrome	3
	742.3	Congenital hydrocephalus	2
	749.0	Cleft palate	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.7	Other deformities of feet	2
	755.0	Polydactyly	2
	755.2	Reduction deformities of upper limb	2
SUFFOLK	752.6	Hypospadias & epispadias	106
	745.5	Ostium secundum atrial septal defect	89
	752.5	Undescended testicle	89
	753.2	Obstructive defects of renal pelvis & ureter	81
	745.4	Ventricular septal defect	75
	747.0	Patent ductus arteriosus	69
	750.5	Congenital hypertrophic pyloric stenosis	36
	747.3	Anomalies of pulmonary artery	30
	755.0	Polydactyly	29
	754.3	Congenital dislocation of hip	27
	758.0	Down syndrome	27
SULLIVAN	745.4	Ventricular septal defect	4
	752.6	Hypospadias & epispadias	4
	750.5	Congenital hypertrophic pyloric stenosis	3
	755.0	Polydactyly	3
	746.7	Hypoplastic left heart syndrome	2
	747.0	Patent ductus arteriosus	2
	752.5	Undescended testicle	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	755.6	Other anomalies of lower limb including pelvic girdle	2
	756.0	Anomalies of skull and face bones	2
TIOGA	752.6	Hypospadias & epispadias	5
	747.3	Anomalies of pulmonary artery	2
	750.5	Congenital hypertrophic pyloric stenosis	2

Section IV - Table 2
Most Frequently Reported Major Malformations By County

Birth Year: 2005

County	ICD-9 Code	Description	Number
TIOGA	273.8	Other disorders of plasma protien	1
	343.9	Infantile cerebral palsy unspecified	1
	741.0	Spina bifida with hydrocephalus	1
	745.5	Ostium secundum atrial septal defect	1
	746.6	Congenital mitral insufficiency	1
	746.7	Hypoplastic left heart syndrome	1
	746.8	Other specified anomalies of heart	1
	749.1	Cleft lip	1
	752.5	Undescended testicle	1
	754.3	Congenital dislocation of hip	1
	755.4	Reduction deformities, unspecified limb	1
	758.0	Down syndrome	1
	759.6	Other hamartoses,nec	1
TOMPKINS	745.4	Ventricular septal defect	4
	753.2	Obstructive defects of renal pelvis & ureter	4
	747.0	Patent ductus arteriosus	2
	752.5	Undescended testicle	2
	758.0	Down syndrome	2
	743.5	Congenital anomalies of posterior segment of eye	1
	744.0	Anomalies of ear causing impairment of hearing	1
	745.5	Ostium secundum atrial septal defect	1
	746.0	Anomalies of pulmonary valve	1
	746.4	Congenital insufficiency of aortic valve	1
	746.7	Hypoplastic left heart syndrome	1
	747.1	Coarctation of aorta	1
	748.5	Agenesis, hypoplasia & dysplasia, lung	1
	749.0	Cleft palate	1
	750.3	Tracheoesophageal fistula, esophageal atresia & stenosis	1
	750.5	Congenital hypertrophic pyloric stenosis	1
	751.1	Atresia & stenosis of small intestine	1
	751.2	Atresia & stenosis of large intestine, rectum, & anal canal	1
	752.6	Hypospadias & epispadias	1
	753.3	Other specified anomalies of kidney	1
	754.3	Congenital dislocation of hip	1
	754.5	Varus deformities of feet	1
	754.6	Valgus deformities of feet	1
	754.8	Other specified nonteratogenic anomalies	1
	755.0	Polydactyly	1
	756.6	Anomalies of diaphragm	1
ULSTER	752.6	Hypospadias & epispadias	12
	750.5	Congenital hypertrophic pyloric stenosis	10
	752.5	Undescended testicle	9
	753.2	Obstructive defects of renal pelvis & ureter	7
	746.8	Other specified anomalies of heart	5

Section IV - Table 2
Most Frequently Reported Major Malformations By County

Birth Year: 2005

County	ICD-9 Code	Description	Number
ULSTER	753.1	Cystic kidney disease	5
	745.4	Ventricular septal defect	4
	745.5	Ostium secundum atrial septal defect	4
	524.0	Major anomalies of jaw size	3
	746.0	Anomalies of pulmonary valve	3
	756.4	Chondrodystrophy	3
WARREN	752.6	Hypospadias & epispadias	4
	745.4	Ventricular septal defect	3
	752.5	Undescended testicle	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	754.3	Congenital dislocation of hip	3
	755.6	Other anomalies of lower limb including pelvic girdle	3
	745.5	Ostium secundum atrial septal defect	2
	746.4	Congenital insufficiency of aortic valve	2
	747.1	Coarctation of aorta	2
	750.5	Congenital hypertrophic pyloric stenosis	2
	753.4	Other specified anomalies of ureter	2
	758.0	Down syndrome	2
WASHINGTON	750.5	Congenital hypertrophic pyloric stenosis	4
	749.0	Cleft palate	3
	752.5	Undescended testicle	3
	753.3	Other specified anomalies of kidney	3
	756.0	Anomalies of skull and face bones	3
	746.4	Congenital insufficiency of aortic valve	2
	752.6	Hypospadias & epispadias	2
	753.2	Obstructive defects of renal pelvis & ureter	2
	754.3	Congenital dislocation of hip	2
	754.5	Varus deformities of feet	2
WAYNE	745.4	Ventricular septal defect	7
	750.5	Congenital hypertrophic pyloric stenosis	5
	752.5	Undescended testicle	5
	752.6	Hypospadias & epispadias	5
	753.2	Obstructive defects of renal pelvis & ureter	5
	754.5	Varus deformities of feet	5
	745.5	Ostium secundum atrial septal defect	4
	747.3	Anomalies of pulmonary artery	4
	742.1	Microcephalus	2
	746.0	Anomalies of pulmonary valve	2
	753.1	Cystic kidney disease	2
	758.0	Down syndrome	2

Section IV - Table 2
Most Frequently Reported Major Malformations By County

Birth Year: 2005

County	ICD-9 Code	Description	Number
WESTCHESTER	752.6	Hypospadias & epispadias	62
	753.2	Obstructive defects of renal pelvis & ureter	49
	745.4	Ventricular septal defect	48
	745.5	Ostium secundum atrial septal defect	48
	752.5	Undescended testicle	41
	755.0	Polydactyly	41
	747.0	Patent ductus arteriosus	32
	750.5	Congenital hypertrophic pyloric stenosis	27
	754.3	Congenital dislocation of hip	15
	754.5	Varus deformities of feet	15
WYOMING	747.0	Patent ductus arteriosus	6
	745.4	Ventricular septal defect	5
	750.5	Congenital hypertrophic pyloric stenosis	3
	753.2	Obstructive defects of renal pelvis & ureter	3
	746.6	Congenital mitral insufficiency	2
	748.5	Agenesis, hypoplasia & dysplasia, lung	2
	751.3	Hirschprung's disease & other functional disorders of colon	2
	752.5	Undescended testicle	2
	754.3	Congenital dislocation of hip	2
	756.7	Anomalies of abdominal wall	2
YATES	753.2	Obstructive defects of renal pelvis & ureter	2
	228.0	Hemangioma, any site	1
	742.4	Other specified anomalies of brain	1
	745.1	Transposition of great vessels	1
	745.5	Ostium secundum atrial septal defect	1
	751.1	Atresia & stenosis of small intestine	1
	752.5	Undescended testicle	1
	753.0	Renal agenesis & dysgenesis	1
	754.3	Congenital dislocation of hip	1
	754.5	Varus deformities of feet	1

Section V
Comparison of Selected Malformation Prevalence with Other Birth Defects Registries

Introduction to Table

The CMR relies on reports from hospitals and physicians for case ascertainment. Underreporting is an obvious concern, and the CMR over the years has developed methods to improve case ascertainment and monitor hospital reporting (Appendix 3). In this section, CMR live birth prevalences are compared with the national prevalence estimates for 21 selected defects developed by the Centers for Disease Control and Prevention (CDC) and the National Birth Defects Prevention Network (NBDPN).[1] The 21 defects were selected as they are generally diagnosed soon after birth and the accuracy of diagnosis should be similar across sites[1].These estimates were based on 11 registries which use active case-finding. Active case-finding uses data collection specialists who go to hospitals to identify and abstract records of children with malformations. The active case-finding systems were chosen as they have similar methodology and prevalence estimates are usually higher in systems using active case finding, although variation was observed even among the 11 active case finding systems (See Figure 1 in Canfield[1)].

As can be seen from Table 1, the CMR prevalences are equal to or higher than the lower boundary of the actual range of the 11 registries for 16 of the 21 defects (bold prevalences) and five of the defects are equal to or higher than the lower 95 percent confidence interval (CI) (boxed prevalences). The prevalences are generally higher for New York State excluding New York City than for New York City (18 defect prevalences are equal to or higher than the lower boundary of the actual range of the 11 registries compared to 12 for New York City).

The interpretation of differences among registry prevalences is difficult. The lower prevalences of the CMR for neural tube defects (NTD), anophthalmia/microphthalmia and trisomy 18 is most likely due to the lack of reports on terminations as termination rates for these conditions are high. The lower prevalence for cleft lip with/without cleft palate is more difficult to explain. There has been little variation in the prevalence since 1983 and it is an easily identified condition. There is also a wide variation within New York State itself from 5.4 in New York City to 13.0 in another region. The lower rates in limb reduction are also more difficult to explain as these are also easily recognizable defects. We have noted that the rate has been declining over several years, especially for lower limbs and plan to examine defect trends in a future report.

None of the 11 registries had all 21 defect prevalences fall within the 95 percent confidence intervals. Several registries would have the highest prevalence for one defect and the lowest prevalence for others. Variation among the registries in the rates of specific defects could reflect demographic differences in the populations as there are racial and ethnic differences in the rates of specific birth defects[1]. The prevalence of Down syndrome, trisomy 18 and trisomy 13 is highly dependent upon the maternal age distribution, age-specific pregnancy rates and women's use of prenatal diagnosis and pregnancy termination. The lower live birth prevalence rates of these chromosomal abnormalities in the CMR may be partially attributable to one or more of these factors. However, the source(s) of much of the variation is unclear and there may be true geographic differences. A comparison of birth defect prevalences between the Metropolitan Atlanta Congenital Defects Program (MACDP) and California Birth Defects Monitoring program (CBDMP) for the years 1983-1988 that adjusted for race, sex and maternal age showed regional differences in arm, hand and limb reduction defects [2.]

CMR staff will continue their efforts to improve reporting (See Appendix 3) and will track our progress using the NBDPN national prevalence estimates.

Section V - Table 1

Prevalence* of Selected Major Birth Defects in New York State
(Birth years: 2003-2005)

Birth Defect Category	New York City	Upstate NY	New York State	NBDPN 1999-2001	95% CI Range
Central nervous system defects					
Anencephalus	0.3	0.5	0.4	2.5	2.3-2.7
Spina bifida without anencephalus	**1.6**	**2.2**	**1.9**	3.7	3.4-3.9
Encephalocele	0.4	**0.6**	**0.5**	0.9	0.8-1.0
Eye defects					
Anophthalmia/ microphthalmia	0.6	**0.9**	**0.8**	2.1	1.9-2.3
Cardiovascular defects					
Common truncus	**1.0**	**0.4**	**0.7**	0.8	0.7-0.9
Transposition of great arteries	**3.6**	**4.7**	**4.2**	4.7	4.5-5.0
Tetralogy of Fallot	**4.8**	**4.9**	**4.9**	3.9	3.8-4.2
Endocardial cushion defect	**2.6**	**3.1**	**2.8**	4.4	4.1-4.6
Hypoplastic left heart syndrome	**2.1**	**2.8**	**2.5**	2.4	2.2-2.6
Orofacial defects					
Cleft palate without cleft lip	**4.2**	**6.5**	**5.4**	6.4	6.1-6.7
Cleft lip with and without cleft palate	5.4	**8.4**	7.0	10.5	10.1-10.9
Gastrointestinal defects					
Esophageal atresia/ tracheosophageal fistula	**2.7**	**2.0**	**2.4**	2.4	2.2-2.6
Rectal and large intestinal atresia/stenosis	**4.4**	**4.7**	**4.6**	4.8	4.5-5.1
Musculoskeletal defects					
Reduction deformity, upper limbs	1.4	**2.1**	1.8	3.8	3.5-4.0
Reduction deformity, lower limbs	0.7	1.0	0.9	1.9	1.7-2.1
Gastroschisis	1.4	**2.5**	**2.0**	3.7	3.5-4.0
Omphalocele	**1.1**	**1.4**	**1.3**	2.1	1.9-2.3
Diaphragmatic hernia	1.8	**2.5**	**2.2**	2.9	2.7-3.1
Chromosomal defects					
Trisomy 13	**0.9**	**0.9**	**0.9**	1.3	1.2-1.5
Down syndrome(trisomy 21)	**11.0**	**13.6**	**12.4**	13.7	13.2-14.1
Trisomy 18	0.9	1.1	1.0	2.4	2.2-2.6

[a] - Prevalence (number of defects per 10,000 live births)

Bold prevalences are within the range of the 11 active registries

Boxed prevalences are equal to or greater than the lower limit of the 95% CI range

References

1. Canfield MA, Honein MA, Yuskiv N et al. National Estimates and Race/Ethnic Specific Variation of Selected Birth Defects in the United States, 1999-2001. Birth Defects Research(Part A) 2006;76:747-756.

2. Schulman J, Edmonds LD, McClern AB, et al. Surveillance for and comparison of birth defect prevelences in two geographic areas - United States 1983-1988. In: CDC Surveillance Summaries; March 19, 1993. *Morbidity and Mortality Weekly Report* 1993; 42(No. SS-1):1-7.

Section VI
Current Topics

Survey of Cleft Palate and Craniofacial Clinics in New York State

Conducted by the Congenital Malformations Registry, 2007-2008

BACKGROUND

The Congenital Malformations Registry (CMR) of the New York State Department of Health (NYSDOH) has been performing surveillance for all major birth defects in children born in New York since 1983. Bound by strict confidentiality laws, the CMR is unable to disclose names of children identified with birth defects. Therefore, when children are reported with birth defects, the CMR staff send the parents of affected newborns informational mailings about potential services that may assist them. In 2004, in response to the *Healthy People 2010* goal [1], the CMR enhanced the mailing for families of children born with oral clefts. Families now receive an introductory letter, a brochure published by the Cleft Palate Foundation titled "Cleft Lip and Cleft Palate – The First Four Years," and a list of craniofacial or oral cleft teams located across New York State (NYS). The list of teams is reviewed periodically for accuracy and completeness and it is not the intention of the CMR to endorse any particular team, but to provide the families with information.

Interdisciplinary approach of caring for children with craniofacial anomalies is recommended by the American Cleft Palate-Craniofacial Association (ACPA).[2] Two national surveys of cleft palate and craniofacial teams were conducted and published in the 1990s. The 1992 report described questionnaire results of responding teams listed in the 1988 ACPA directory and the 1998 paper discussed results of the ACPA self-assessments performed by teams in 1996. The reports provided an overview of the status and makeup of interdisciplinary teams in the United States.[3, 4] Our interest was to obtain information about the cleft palate and craniofacial teams located in New York, a large state with geographically diverse areas and approximately 300-350 cases of orofacial clefts among all births each year.[5] This paper describes an overview of the responses to a written questionnaire sent to New York team leaders in an effort to obtain more information about the teams, their patient populations, medical care they were able to dispense, and barriers they faced.

METHODS

A written questionnaire was developed that inquired as to the team designation, patient population in terms of numbers and ages, referral activities, specialists serving on the team, barriers to care the children received, and support provided to parents, families, and other health care professionals. The survey was sent to the leaders of the teams located in New York. If no response was received, contact was made by email; otherwise, a second contact was made by mail. A third attempt to encourage participation of non-responders was attempted by mail that was addressed to both the team leader and the team coordinator. Descriptive analysis was performed to provide an overview of the responses.

RESULTS

Team Descriptions Eighteen teams were identified in New York State and 15 responding teams included seven of the ten in New York City or Long Island and all of the teams located in the rest of New York. Of the responding teams, three teams were established in the 1950s, nine were founded between 1970 and 1994, two began treating patients since 2000, and one did not provide a year of establishment. When asked which designation applied to their team, six responders selected 'cleft palate team,' two responders selected 'craniofacial team,' and seven responders selected 'both.' One institution that has 'both' designations noted that they have two separate teams (a cleft team and a craniofacial team) and reported some results separately for both teams.

All but one team indicated that they had a designated team coordinator on staff. Team coordinators included the following: nurses, nurse practitioners, speech pathologists, physicians, surgeons, or individuals with college degrees (B.S. or A.S.). Eleven teams offered continuing education opportunities to the team members, and 11 provided continuing education to non-team members. The teams were asked how often they usually meet face-to-face each year. The range of responses was from 6 to 40 times with a median number of 12 meetings per year. All teams said that they routinely formed treatment plans for each patient.

Patient Volumes The approximate number of patients seen per year ranged from 30 to 500 with a median of 150. When asked how many new patients were evaluated each year, the responses ranged from 5 to 200 with a median of 40. All responding teams cared for children (age 0-17 years old), eleven attended to patients 18-35 years old, and 8 teams saw adults over age 35.

Referral Activities Teams reported that patients were generally referred to them from birth hospitals, primary care physicians or pediatricians, geneticists, obstetricians, and surgeons. When asked to comment on issues or problems they encountered regarding referral activities, a few noted that community caregivers (hospitals or private physicians) did not understand the importance of the teams or they were not consistent in their referral patterns. One commented on the problem of insurance companies requiring referrals for each team member rather than for the team as a unit.

Services Provided Each team was asked about the disciplines or specialists who provided services as part of their team (Table 1). They were also asked which disciplines or specialists they would add if the means were available. Three indicated they would not add any other services if they could. Fourteen teams included an otolaryngolist, an orthodontist and a speech pathologist, and 13 had a plastic surgeon serving on their team. Only one team did not have either a pediatric or a general dentist on their team; they referred patients to both disciplines and they would add a pediatric dentist if they had the means. Eleven responders noted that a geneticist served on their team. Regarding psychological services, nine teams included a psychologist, a clinical social worker, or both. Of the six teams that did not have either a psychologist or a social worker, three referred patients to both disciplines and all six would add a psychologist or a social worker if they could. When asked about limitations to the services they provided, nine indicated they were limited by factors such as monetary/budgetary (n=8), staffing (n=7), expertise (n=2), hospital management decisions (n=3), and insurance restrictions (n=2).

Table 1. Number who responded that the discipline served on their team and number who would add the discipline if possible

Discipline/specialist	Served on team	Would add to team
Otolaryngolist	14	0
Orthodontist	14	0
Speech Pathologist	14	0
Plastic surgeon	13	1
Pediatric dentist	13	1
Geneticist	11	2
Neurosurgeon	11	0
Audiologist	10	1
Prosthodontist	9	2
Clinical social worker	8	2
Ophthalmologist	8	1
Radiologist	7	0
Nurse practitioner/Nurse specialist	6	4
General dentist	6	0
Psychologist	5	5

Newborn Care and Family Support Twelve of the 15 responded that their team visited newborns in the birth hospital before the child was discharged, and 11 felt that the parents received appropriate information, counseling and support from a team member if the affected child was born in an affiliated hospital. When asked if they felt parents received appropriate information, counseling and support if a child was born in a hospital in their region that was not affiliated with their team, only three responded "yes". Fourteen teams had a nurse or other professional available to provide counseling or instruction to parents regarding newborn feeding or developmental issues.

Because many families must travel when their children receive care or surgery, teams were asked about support given in terms of arrangements for lodging. Eleven of the teams indicated that arrangements were available for families (i.e., hospital rooms, local hotels, hospital affiliated hotels or apartments, and Ronald McDonald Houses).

Support groups and parent networks can be very important sources of information and comfort to parents of children born with craniofacial malformations. Eleven of the 15 teams said they were aware of community support groups or parent networks in their area available to families; however, only three teams sponsored any support groups or parent networks.

Barriers to care and concerns Survey recipients were asked to consider and rank a list of possible barriers to care for families. Eleven placed insurance issues (no insurance, insurances that did not adequately cover services, or insurances that denied coverage for services) as the barriers seen most often. Other barriers noted were geography, transportation, and language issues. Overall, they had concerns about affected children not receiving the appropriate care for reasons that were beyond the control/scope of their team. Of the 15 teams, 12 said they had concerns and described their concerns as: related to insurance issues (n=6), social – the parents lacked the understanding or financial resources to bring children to appointments (n=2), lack of referrals to teams (n=2), geography issues (n=1), and limited therapy in schools (n=1).

Finally, the survey recipients were asked to comment on any issues raised by the survey, or any other thoughts they wanted to share regarding treatment of children in New York State with craniofacial anomalies. Six comments related to medical insurance, specifically low reimbursement for care or denial for important services such as orthodontics and psychosocial care, especially for children as they enter the teen years. Other issues important to the responders included lack of institutional support for the team, problems related to referrals, laws they viewed as barriers to genetic testing, and the need for more consolidation of care to larger centers with comprehensive capabilities for pediatric care.

DISCUSSION

New York is a very diverse state with population sizes that vary drastically among its 62 counties. Ten of the 18 teams are located in New York City or Long Island, while the remaining eight are located in cities across the State (Buffalo, Rochester, Syracuse, Elmira, Binghamton, Schenectady, Albany and Valhalla). The "upstate" cities vary in size, which may account for the large range noted for numbers of patients seen by each team. While the regions containing small to larger cities have organized craniofacial care teams, there are many areas in New York that are quite rural and distant from the existing teams. However, geography and transportation were not considered high on the list of noted barriers to care.

What did seem to be of concern to most respondents were issues related to insurance coverage. It is likely that team staff spend large amounts of time trying to encourage insurance companies to cover expenses for services and care of children that the medical community deem vital. Insurance coverage for care and treatment of children into the teen years and continuing into adulthood is important to the overall well being of each individual born with a craniofacial anomaly, and may play a significant role in individual's future well-being.

Reimbursement for services may have an impact on medical professionals who choose specialties that are important for the formation and success of interdisciplinary teams. Not all of the New York State teams have staff that encompass all the disciples recommended by the ACPA. Two-thirds of the respondents who said they felt limited to the services they could provide sited staffing issues as a problem. The ACPA recognizes psychological services as an integral component in interdisciplinary team care [2]; however, six responding teams did not have a psychologist or clinical social worker on staff to monitor the development of the children or to support the families.

New York State has 18 organized care teams identified by CMR staff; however, there may be other organized teams that were not identified. Furthermore, there may be children with craniofacial anomalies who are followed by physicians and surgeons who are very dedicated to the well-being of their patients but who do not work as a member of a team. While it is recommended that affected children be enrolled in interdisciplinary team care settings, ultimately it is the quality of care they receive that is important. Measuring outcomes of treatment and

services received by children is a difficult undertaking. CMR staff are collaborating on several projects with researchers in Iowa, Arkansas and New York City to study the access to medical care and outcomes experienced by children affected by selected craniofacial malformations. The collaborative efforts also include a survey of primary care physicians and pediatricians in New York, Iowa and Arkansas to obtain information about their experiences in caring for children with orofacial clefts. The survey of craniofacial and cleft care teams in New York described here, in addition to the information drawn from the other research projects mentioned might allow for a more complete picture of the care available to children and their families of New York who are affected by craniofacial anomalies.

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APPENDICES

Appendix 1

Classification of Codes

Congenital malformations have traditionally been divided into categories of "major" and "minor". A major anomaly has an adverse effect on the individual's health, functioning or social acceptability. A minor anomaly is generally considered of limited social or medical significance. While minor anomalies in themselves do not greatly affect the child, they can be related to major anomalies or be indications of certain syndromes.[1,2]

The division between major and minor is far from perfect. No standard lists or definitions exist. We used several sources, including the practices of other registries, to develop a list of minor anomalies.[3, 4, 5] One serious problem in making this distinction is that some ICD-9-CM codes include major and minor malformations under the same code. A more specific coding scheme that eliminates most of these problems has been adopted.

Following is a general listing of conditions included in this report and their classification. A few codes are not listed since they contain only a very few cases. Reporting hospitals receive a CMR Handbook with a complete, detailed list of reportable anomalies.

Major Malformations

658.8	Amniotic Bands
740 - 759*	Congenital Anomalies
760.71	Fetal Alcohol Syndrome
771.0 - 771.2	Congenital Infections: including rubella, cytomegalovirus toxoplasmosis and herpes simplex

***See list of minor and excluded codes**

Minor Malformations

214	Lipoma
216	Benign neoplasm of skin
228.01	Hemangioma of skin
550	Inguinal hernia in males
553.1	Umbilical hernia
743.65	Specified congenital anomalies of lacrimal passages
744.1	Accessory auricle
744.29	Other specified anomalies of ear
744.3	Unspecified anomaly of ear
744.4	Branchial cleft cyst
744.89	Other specified anomalies of face and neck
744.9	Other unspecified anomalies of face and neck
747.0	Patent ductus arteriosis, if birth weight <1500 grams
747.5	Single umbilical artery
752.41	Embryonic cyst of cervix, vagina and external female genitalia
752.42	Imperforate hymen
752.5	Undescended testicle, if birth weight < 2500 grams
754.61	Congenital pes planus
755.0	Polydactyly
755.11, 755.13	Syndactyly without fusion of bone
757.2	Dermatoglyphic anomalies
757.32	Vascular hamartomas
757.33	Congenital pigmentation anomalies of skin
757.39	Other anomalies of skin
757.4	Specified anomalies of hair
757.5	Specified anomalies of nails
757.6	Specified anomalies of breast
757.8	Other specified anomalies of integument
757.9	Unspecified anomalies of the integument

Exclusions

750.0	Tongue tie
758.4	Balanced autosomal translocation in normal individual
778.6	Congenital hydrocele

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Appendix 2

Birth Certificate Matching

Birth certificate matching is a vital part of registry activities. This serves to verify the individual's identity and distinguish him or her from all others and provides additional information about the baby and the mother. The matching is used to determine maternal residence at birth and to verify race and birth weight. Matched cases provide a basis to calculate population-based rates. It is critical to match a high percentage of cases to calculate rates accurately and to conduct meaningful surveillance.

Birth certificate matching is carried out by a computer program that compares the birth certificate records for a given year to the CMR file of cases who were born in that year. A deterministic matching method is applied to identify all possible matches, using combinations of identifying variables such as name, date of birth, medical record number and mother's name and address information. Matching scores are assigned to each criterion. Assigning different points to different identifiers provides a way to recognize variations in quality or reliability of different data items. The records are compared on identifying variables that are available until (1) a match is found, (2) a possible match is found or (3) the list is exhausted without finding a match. Possible matches are reviewed by CMR staff and a decision made about whether there is a match.

The matching process is repeated until about 95 percent of reported cases are matched. This is a compromise between completeness and efficiency. After about 90 percent of cases are matched, each additional percentage requires greater and greater effort. The ability to review a copy of the birth certificate greatly enhances the chance of making a match. Matching is more complete for cases born in the state outside New York City than for New York City cases.

Appendix 3

Case Ascertainments and Data Quality Assurance

The CMR uses the method of passive case ascertainment of birth defects that occur among live births, with an active follow-up for assuring the accuracy and completeness of case reporting. Birth defect cases reported from hospitals and physicians are reviewed and the diagnoses are coded by the registry's trained staff. Reporting hospitals and physicians are contacted for cases that have insufficient diagnostic information for coding. CMR staff recognizes that completeness, accuracy and timeliness are the hallmarks of a good surveillance system. However, these attributes exist in tension, "conflicting principles" (Kallen 1988). Steps taken to improve completeness and accuracy may actually reduce timeliness. From the very beginning, the CMR has built in procedures to improve the quality of the data in the CMR. These systems have changed over time (Sekhobo and Druschel 2001; Druschel et al, 2001) and the CMR now has three major approaches to improving data quality: 1) matching to hospital discharge data, the Statewide Planning and Research Cooperative System (SPARCS) for completeness; 2) the web-based reporting system, the Health Provider Network (HPN) for timeliness and completeness; 3) on-site hospital audits for completeness and accuracy. In addition, we also periodically request medical records and compare them to the hospital's report for an additional review of accuracy.

SPARCS Audits For the SPARCS audit, children age 2 years or younger and diagnosed with reportable birth defects are selected from SPARCS files of all reporting hospitals and matched to the CMR database for the same birth year period. As about 90 percent of children reported to the CMR were diagnosed in the first six months of life, CMR staff begin to audit hospitals 12 to 24 months after the reporting period for each year of birth. Unmatched reports from the SPARCS hospital discharge files are sent to the hospital, requesting submission of the missed reports. A recent study (Wang et al, 2005) demonstrated that using hospital discharge data to improve case ascertainment is a valuable and effective method of enhancing birth defect surveillance, particularly for those hospitals with low reporting rates. Hospital audits resulted in not only added new reports (comprised 21.4 percent of all CMR reports) to the CMR but also improved reporting for subsequent years, probably due to hospitals' positively reacting to the audits. Auditing hospitals by CMR staff sent a message to reporting hospitals that both the quality and the quantity of their reports are closely monitored.

HPN Reporting A web-based reporting, data management and communication system has been successfully developed and implemented by CMR staff (Wang et al, 2007a, Steen et al, 2008). After pilot testing with two hospitals in 2001, the system was phased in for reporting in 2003. By January 2006, the CMR had converted all reporting hospitals statewide from a manual, paper-based reporting system to the web-based system. This new system provides a platform-independent environment for data submission, retrieval and analysis and offers a secure, cost-effective solution for participating hospitals. An authorized user can submit/edit data and view, update or query their case information dynamically from the CMR's database using any personal computer equipped with an internet browser from any geographic area throughout the state. This innovative system enables CMR staff to review and perform quality assurance on every report submitted and to query hospitals quickly about submitted reports. A study that evaluated the completeness of submitted case information and timeliness of reporting to the CMR and the effectiveness of the HPN communication and query system when compared to the previous manual, paper-based system found that the implementation of the HPN system has resulted in more timely submission of cases and promoted effective communication between the CMR and reporting hospitals. There was a nearly 50 percent reduction in median days used for reporting. (Wang et al, 2007b).

Monitoring Hospital Reporting CMR staff have developed on-line SAS/IntrNet applications which empower the users to search and retrieve hospital submitted cases, generate real-time reports and perform simple statistical analysis using the CMR's database (Wang et al, 2008). For instance, CMR staff can select a reporting hospital and discharge years of interest and then, generate a real-time report table which lists the number of cases by discharge year and month. By reviewing this report, CMR staff are able to identify hospitals with unusual reporting patterns or problems, for instance, if they stopped or skipped reporting for certain months or years.

On-site Hospital Audits On-site hospital audits began in August of 2003 as an additional surveillance tool. CMR staff needed to know if all malformations were being captured from medical records, and if the reports were complete and accurate. This was piloted in 2002 and implemented in 2003. The procedure begins when the CMR announces to the hospital that they will be making an "in-house chart review or audit" and requests the hospital in question to send a discharge summary for all children 2 years of age and younger for a specific discharge period, usually one year. The list includes all children discharged in that given year, not just those with a congenital code. This is done so that reportable conditions that may have been miscoded can be identified. CMR staff review the discharge list, comparing it to the list of children who have already been reported to the CMR. A list of reported, not reported and partially reported cases is made. Depending on the time frame and number of auditors available, the entire list or a subset of this list will be sent to the hospital and they will be requested to produce the charts so that CMR staff can review them. CMR staff will spend between 1 and 2 days at a facility reviewing records. At the completion of the review, the facility will be asked to report any case that is considered by the CMR staff as reportable but not previously reported as well as any partially reported cases that need to be completed. A written summary of the audit findings is sent to the Director of Health Information Management including comments that may indicate what chronic reporting problems were evident. Since 2003, 62 hospitals have had an "in-house" audit; 3598 charts have been reviewed; 1311 cases that were not previously reported were flagged and subsequently reported, 326 cases that were partially reported were completed and 154 cases with incorrect diagnoses reported were corrected or deleted.

Hospital Report Card In order to improve the completeness of case reporting and the accuracy of reported cases, CMR staff have developed an on-line application to generate report cards for hospitals to track their reporting progress in 2008. The first report card summarizing reporting status and progress of hospitals for the reporting period of June 1 - December 31, 2007 was sent to each individual hospital in April 2008. The report cards for all reporting hospitals are generated bi-annually and made available online for the hospital officials.

Summary Surveillance requires on-going efforts to respond to changes in resources and technologies. There must also be constant communication and feedback between the reporting sources and the surveillance system. The CMR has developed several methods to monitor and improve the system's completeness, accuracy and timeliness. CMR staff recognize that as a 'passive' reporting system much additional work must be done to be able to provide data of good quality. While 'active'case ascertainment systems seem to provide more completeness and accuracy, they require much higher funding levels and many more staff. In this era of cutbacks, these funding levels can be difficult to maintain and some of these systems have been forced to reduce their activities or decrease their areas of coverage. The CMR has seen many staff reductions over the years but by making use of new technologies has been able to improve the system. However, further improvements are needed and the CMR will continue to review procedures and develop new methods. The CMR is currently investigating ways to use hospital discharge summaries (most of which are electronic) as an additional source of case finding. As

more and more hospitals go to electronic medical records, these might also assist us in case finding and confirmation of diagnoses. Birth defects are a serious health issue for affected infants and children and their families. With so many different conditions, surveillance of birth defects can be challenging but must be done so that they can be tracked and studied.

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Appendix 4

BPA Codes

Many birth defects registries use a coding system modified from the British Pediatric Association (BPA). This coding system provides more specificity than the ICD-9 system. The Centers for Disease Control and Prevention Metropolitan Atlanta Congenital Defects Program (MACDP) has developed codes that group conditions. The table below shows the MACDP codes and the corresponding BPA and ICD-9 codes. The ICD-9 code may include conditions others than those specified by the BPA code. For example, ICD-9 code 756.7 includes both gastroschisis and omphalocele, but the BPA code allows these conditions to be distinguished.

MACDP Code	Condition	ICD-9	BPA Code
CENTRAL NERVOUS SYSTEM			
A01	Anencephaly	740.0, 740.1, 740.2	740.00, 740.01, 740.02, 740.03, 740.08, 740.10, 740.20, 740.21, 740.29
A02	Spina Bifida with Hydrocephaly	741.00, 741.01, 741.02, 741.03	741.000, 741.001, 741.002, 741.003, 741.004, 741.008, 741.009, 741.011, 741.012, 741.013, 741.014, 741.018, 741.019, 741.021, 741.022, 741.023, 741.024, 741.028, 741.029-741.599
A03	Spina Bifida without Hydrocephaly	741.90, 741.91, 741.92, 741.93	741.701, 741.702, 741.703, 741.704, 741.708, 741.709- 741.999
A13	Encephalocele	742.0	742.000, 742.080, 742.085, 742.086, 742.090
A15	Hydrocephaly	742.3	742.300, 742.310, 742.320, 742.380, 742.390
A16	Microcephalus	742.1	742.100, 742.150
EYE / EAR			
B01	Anophthalmia, Microphthalmia	743.00, 743.10, 743.11, 743.12	743.0000, 743.1000, 743.1009, 743.0003, 743.0006, 743.1001, 743.1002
B03	Glaucoma	743.20, 743.21, 743.22	743.2000, 743.210, 743.2001, 743.220
B04	Cataract	743.30, 743.31, 743.32, 743.33, 743.34, 743.35, 743.36, 743.37, 743.39	743.320, 743.325, 743.3261, 743.3262, 743.3263, 743.3264, 743.300, 743.310, 743.340, 743.3806, 743.330, 743.3269, 743.3809, 743.390
B54	Ear anomaly with hearing loss	744.00, 744.01, 744.02, 744.03, 744.04, 744.05, 744.09	744.0001, 744.0101, 744.0002, 744.0902, 744.0203, 744.0204 744.030, 744.0109, 744.0900
CARDIAC			
D01	Truncus arteriosus	745.0	745.000, 745.010
D02	Transposition of great vessels	745.10, 745.11, 745.12, 745.19	745.1001, 745.110, 745.1801, 745.120, 745.1809, 745.190
D03	Tetralogy of Fallot	745.2	745.200, 745.210
D04	Single ventricle	745.3	745.300
D05	VSD	745.4	745.480, 745.485, 745.486, 745.487, 745.490
D52	Hypoplastic left heart	746.7	746.700
D53	Total anomalous pulmonary venous return	747.41	747.420
RESPIRATORY			
E01	Choanal atresia	748.0	748.000
E06	Agenesis of lung	748.5	748.500, 748.510, 748.520, 748.580, 748.590

MACDP Code	Condition	ICD-9	BPA Code
CLEFTS			
F01	Cleft palate	749.00, 749.01, 749.02, 749.03, 749.04	749.010, 749.020, 749.030, 749.050, 749.060, 749.070, 749.090, 749.001, 749.002, 749.003, 749.041, 749.042, 749.043, 749.080
F02	Cleft lip with or without cleft palate	749.10, 749.11, 749.12, 749.13, 749.14, 749.20, 749.21, 749.22, 749.23, 749.24, 749.25	749.1010, 749.1020, 749.1030, 749.1100, 749.120, 749.1901, 749.1011, 749.1021, 749.1031, 749.1012, 749.1022, 749.1032, 749.1103, 749.1104, 749.2900, 749.2011, 749.2021, 749.2031, 749.2012, 749.2022, 749.2032, 749.2103, 749.2104, 749.2015, 749.2025, 749.2035, 749.2105, 749.2203, 749.2905
GASTRO-INTESTINAL			
F14	Stenosis or atresia of duodenum	751.1	751.100
F15	Other stenosis or atresia of small intestine	751.1	751.110, 751.120, 751.190, 751.195
F16	Stenosis or atresia of rectum or anus	751.2	751.210, 751.220, 751.230, 751.240
F17	Hirschsprung's Disease	751.3	751.300, 751.310, 751.320, 751.303
F18	Malrotation of intestine	751.4	751.400, 751.410, 751.420, 751.490, 751.495
F21	Biliary atresia	751.61	751.6501
GENITO-URINARY			
H01	Renal agenesis	753.0	753.000, 753.009, 753.010
H06	Obstruction of kidney or ureter	753.20, 753.21, 753.22	753.220, 753.221, 753.240, 753.241, 753.242, 753.243, 753.244, 753.290, 753.299
H09	Bladder or urethra obstruction	753.6	753.600, 753.610, 753.620, 753.630, 753.690
MUSCULOSKELETAL			
J02	Curvature of spine (scoliosis or lordosis)	754.2	754.200, 754.210, 754.220
J03	Dislocation of hip	754.30, 754.31	754.3000, 754.3010, 754.3020, 754.3030
J11	Arthrogryposis multiplex congenita	754.89	755.800
K01	Reduction deformity - upper limb	755.20, 755.21, 755.22, 755.23, 755.24, 755.25, 755.26, 755.27, 755.28, 755.29	755.200, 755.230, 755.240, 755.2901, 755.5851, 755.2602, 755.265, 755.2702, 755.280, 755.2902, 755.210, 755.218, 755.220, 755.2606, 755.2707, 755.2801, 755.247, 755.2609, 755.2709, 755.2900, 755.5800, 755.5850, 755.59859
K02	Reduction deformity - lower limb	755.30, 755.31, 755.32, 755.33, 755.34, 755.35, 755.36, 755.37, 755.38, 755.39	755.300, 755.330, 755.3401, 755.33901, 755.6851, 755.360, 755.380, 755.3103, 755.3104, 755.318, 755.3801, 755.320, 755.365, 755.366, 755.3802, 755.3409, 755.3900, 755.6859
K05	Amniotic bands	658.80	658.801
N01	Diaphragmatic hernia	756.6	756.610, 756.615, 756.616
N02	Omphalocele	756.79	756.700
N04	Gastroschisis	756.79	756.710
SYNDROMES			
R01	Down Syndrome	758.0	758.000, 758.010, 758.020, 758.030, 758.040, 758.050, 758.09
R02	Patau Syndrome (Trisomy 13)	758.1	758.100, 758.110, 758.120, 758.130, 758.140, 758.150, 758.190
R03	Edwards Syndrome (Trisomy 18)	758.2	758.200, 758.210, 758.220, 758.230, 758.290, 758.295, 758.296
S02	Fetal Alcohol Syndrome	760.71	760.710, 760.715, 760.718
W03	Conjoined twins	759.4	759.400, 759.410, 759.420, 759.430, 759.440, 759.480, 759.490

Appendix 5

Glossary of Birth Defects and Related Terms

(Courtesy of the Texas Birth Defects Monitoring Division, August 2008)

Agenesis Absence of part(s) of the body.

Agenesis, aplasia, or hypoplasia of the lung The absence or incomplete development of a lung or lung tissue.

Anencephaly Congenital absence of the skull, with cerebral hemispheres completely missing or reduced to small masses attached to the base of the skull. Anencephaly is not compatible with life.

Aniridia The complete absence of the iris of the eye or a defect of the iris. Can be congenital or traumatically induced.

Anomalies of the tricuspid valve Includes tricuspid valve atresia or stenosis, as well as enlargement, dilation, or aneurysm of the tricuspid valve. See also tricuspid valve atresia or stenosis.

Anophthalmia A developmental defect characterized by complete absence of the eyes, or by the presence of vestigial eyes.

Anotia A congenital absence of one or both ears.

Aorta The large arterial trunk that carries blood from the heart to be distributed by branch arteries through the body

Aortic valve stenosis A cardiac anomaly characterized by a narrowing or stricture of the aortic valve. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. This condition can be repaired surgically in some cases.

Atresia Imperforation; absence or closure of a normal opening.

Atrial septal defect A congenital cardiac malformation in which there are one or several openings in the atrial septum (muscular and fibrous wall between the right and left atria) allowing a mixing of oxygenated and unoxygenated blood. The openings vary in size and may resolve without treatment or may require surgical treatment. Also called *ostium secundum defect.*

Atrium One of the two upper chambers of the heart (plural atria). The right atrium receives unoxygenated blood from the body. The left atrium receives oxygenated blood from the lungs.

Biliary atresia A congenital absence or underdevelopment of one or more of the ducts in the biliary tract. Correctable surgically.

Birth prevalence

$$\frac{\text{\# of cases with birth defect A in an area and time period}}{\text{\# of live births in that area and time period}} \times 10{,}000$$

Bladder exstrophy Incomplete closure of the anterior wall of the bladder and the abdominal cavity. The upper urinary tract is generally normal. Often associated with anorectal and genital malformations, and epispadias. Affected persons are at a markedly increased risk of bladder carcinoma (squamous cell). This condition is usually corrected surgically after birth.

Cataract An opacity (clouding) of the lens of the eye.

Choanal atresia or stenosis A congenital anomaly in which a bony or membranous formation blocks the passageway between the nose and the pharynx. This defect is usually repaired surgically after birth. Bilateral choanal atresia is a surgical emergency.

Cleft lip The congenital failure of the fetal components of the lip to fuse or join, forming a groove or fissure in the lip. Infants with this condition can have difficulty feeding, and may use assistive devices for feeding. This condition is corrected when the infant can tolerate surgery.

Cleft palate The congenital failure of the palate to fuse properly, forming a grooved depression or fissure in the roof of the mouth. This defect varies in degree of severity. The fissure can extend into the hard and soft palate and into the nasal

cavities. Infants with this condition have difficulty feeding, and may use assistive devices for feeding. Surgical correction is begun as soon as possible. Children with cleft palates are at high risk for hearing problems due to ear infections.

Cluster An apparently unusual concentration of a health condition in a particular area and time period.

Coarctation of the aorta Localized narrowing of the aorta. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. This condition can vary from mild to severe. Surgical correction is recommended even for mild defects.

Common truncus ateriosus A congenital heart defect in which the common arterial trunk fails to divide into pulmonary artery and aorta. This is corrected surgically.

Confidence interval (95 percent) The interval that contains the true prevalence (which we can only estimate) 95 percent of the time. See Methods for more explanation.

Congenital Existing at or dating from birth.

Congenital hip dislocation A congenital defect in which the head of the femur does not articulate with the acetabulum of the pelvis because of an abnormal shallowness of the acetabulum. Treatment in early infancy consists of bracing of the joint to cause a deepening of the acetabulum.

Craniosynostosis A premature ossification (closing) of the cranial sutures before birth or soon after birth. This condition is occasionally associated with other skeletal defects. If no surgical correction is made, the growth of the skull is inhibited, and the head is deformed. The eyes and the brain are often damaged.

Diaphragmatic hernia A failure of the diaphragm to form completely, leaving a hole. Abdominal organs can protrude through the hole into the chest cavity and interfere with development of the heart and lungs. Usually life-threatening and requires emergent surgery.

Down syndrome (Trisomy 21) The chromosomal abnormality characterized by an extra copy of chromosome 21. In rare cases this syndrome is caused by translocation. The extra copy can be free-lying, or can be attached to some other chromosome, most frequently number 14. Down syndrome can occur in mosaic, so that there is a population of normal cells and a population of trisomy 21 cells. Down syndrome is characterized by moderate to severe mental retardation, sloping forehead, small ear canals, flat bridged nose, and short fingers and toes. One third of infants have congenital heart disease, and one third have duodenal atresia. (Both can be present in the same infant.) Affected people can survive to middle or old age. There is an increased incidence of Alzheimer disease in adults with Down syndrome.

Dysgenesis Impaired or faulty development of part(s) of the body.

Ebstein anomaly A congenital heart defect in which the tricuspid valve is displaced downward into the right ventricle causing abnormal patterns of cardiac circulation.

Edwards syndrome (Trisomy 18) The chromosomal abnormality characterized by an extra copy of chromosome 18. The extra chromosome can be free lying or attached to another chromosome. Trisomy 18 can occur in mosaic. Edwards syndrome is characterized by mental retardation, neonatal hepatitis, low-set ears, skull malformation, and short digits. Cardiac and renal anomalies are also common. Survival for more than a few months is rare.

Embryogenesis The development and growth of an embryo, especially the period from the second through the eighth week after conception.
Encephalocele The protrusion of the brain substance through a defect in the skull.

Endocardial cushion defect A variety of septal defects (malformations of the walls separating the two atria and two ventricles of the heart) resulting from imperfect fusion of the endocardial cushions in the embryonic heart.

Epispadias A congenital defect in which the urinary meatus (urinary outlet) opens above (dorsal to) the normal position. The urinary sphincters are defective, so incontinence does occur. Surgical correction is aimed at correcting incontinence and permitting sexual functioning. The corresponding defect in females is rare. *See also Hypospadias.*

Esophageal stenosis or atresia A narrowing or incomplete formation of the esophagus. Usually a surgical emergency. Frequently associated with a tracheoesophageal fistula.

Fetal alcohol syndrome A constellation of physical abnormalities (including characteristic abnormal facial features and growth retardation), and problems of behavior and cognition in children born to mothers who drank alcohol during pregnancy.

Fistula An abnormal passage from an internal organ to the body surface or between two internal organs or structures.

Folate B vitamin necessary for red blood cell production; folate deficiency can lead to anemia and, during embryogenesis, can affect the normal development of the fetus' neural tube; found in liver, green leafy vegetables, beans, beets, broccoli, cauliflower, citrus fruits, and sweet potatoes. *See folic acid.*

Folic acid One of the B vitamins especially important for a woman to take before conception to help prevent neural tube defects in a fetus; essential for DNA synthesis and therefore the growth and division of cells; obtained from fortified foods or from a multivitamin containing at least 4mg; also found in natural sources including liver, beans, and leafy green vegetables. While folate and folic acid are both forms of water-soluble B vitamins, folic acid refers to the synthetic vitamin used in supplements, whereas folate is the form found in foods.

Gastroschisis A congenital opening of the abdominal wall with protrusion of the intestines. This condition is surgically treated. Contrast with Omphalocele, below.

Hernia A protrusion of an organ or part through connective tissue or through a wall of the cavity in which it is normally enclosed.

Hirschsprung disease The congenital absence of autonomic ganglia (nerves controlling involuntary and reflexive movement) in the muscles of the colon. This results in immobility of the intestines and may cause obstruction or stretching of the intestines. This condition is repaired surgically in early childhood by the removal of the affected portion of the intestine.

Holoprosencephaly Failure of the brain to develop into two equal halves, so there is structural abnormality of the brain. There may be associated midline facial defects including cyclopia (fusion of the eye orbits into a single cavity containing one eye) in severe cases. About half the cases are probably due to a single gene defect (the HPE gene). Frequently occurs with Trisomy 13.

Hydrocephaly The abnormal accumulation of fluid within the spaces of the brain.

Hyperplasia Overgrowth characterize by an increase in the number of cells of a tissue.

Hypoplasia A condition of arrested development in which an organ or part remains below the normal size or in an immature state.

Hypoplastic left heart syndrome Atresia, or marked hypoplasia, of the aortic opening or valve, with hypoplasia of the ascending aorta and defective development of the left ventricle (with mitral valve atresia). This condition can be surgically repaired in a series of three procedures over a period of one year. Transplantation is also a treatment. This condition is usually fatal in the first month of life if not treated.

Hypospadias A congenital defect in which the urinary meatus (urinary outlet) is on the underside of the penis or on the perineum (area between the genitals and the anus). The urinary sphincters are not defective so incontinence does not occur. The condition may be surgically corrected if needed for cosmetic, urologic, or reproductive reasons. The corresponding defect in women is rare. *See also epispadias*

Limb defects See Reduction defects.

Meninges Membranes that cover the brain and spinal cord.

Microcephaly The congenital smallness of the head, with corresponding smallness of the brain.

Microphthalmia The congenital abnormal smallness of one or both eyes. Can occur in the presence of other ocular defects.

Microtia A small or maldeveloped external ear and atretic or stenotic external auditory canal.

Mosaic In genetics, this refers to an individual organism that has two or more kinds of genetically different cell types. The degree of abnormality depends on the type of tissue containing affected cells. Individuals may vary from near normal to full manifestation of the genetic syndrome. Can occur in any chromosome abnormality syndrome.

Neural tube defect A defect resulting from failure of the neural tube to close in the first month of pregnancy. The major conditions include anencephaly, spina bifida, and encephalocele.

Obstructive genitourinary defect Stenosis or atresia of the urinary tract at any level. Severity of the defect depends largely upon the level of the obstruction. Urine accumulates behind the obstruction and damages the organs.

Omphalocele The protrusion of an organ into the umbilicus. The defect is usually closed surgically soon after birth. Contrast with Gastroschisis.

Ostium secundum defect See atrial septal defect.

Patau syndrome (Trisomy 13) The chromosomal abnormality caused by an extra chromosome 13. The extra copy can be free-lying, or can be attached to some other chromosome. Patau syndrome can occur in mosaic so that there is a population of normal cells and a population of trisomy 13 cells. Patau syndrome is characterized by impaired midline facial development, cleft lip and palate, polydactyly, and mental retardation. Most infants do not survive beyond 6 months of life.

Patent ductus arteriosus A blood vessel between the pulmonary artery and the aorta. This is normal in fetal life, but can cause problems after birth, particularly in premature infants. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. The vast majority close spontaneously and cause no problems. Medical or surgical correction may be done. This is only an abnormality if it causes significant medical problems.

Poisson regression A type of statistical analysis based on the Poisson distribution used to compare rates of rare occurrences such as birth defects between different population groups, different areas, or different times.

Prevalence With respect to the prevalence of birth defects, see "*Birth prevalence*".

Pulmonary artery anomaly Abnormality in the formation of the pulmonary artery such as stenosis or atresia. See also common truncus.

Pulmonary valve atresia or stenosis A congenital heart condition characterized by absence or constriction of the pulmonary valve. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. This condition can vary from mild to severe. Mild forms are relatively well tolerated and require no intervention. More severe forms are surgically corrected.

Pyloric stenosis A narrowing of the pyloric sphincter at the outlet of the stomach. This causes a blockage of food from the stomach into the small intestine. Usually treated surgically.

Reduction defects of the lower limbs The congenital absence of a portion of the lower limb. There are two general types of defect, transverse and longitudinal. Transverse defects appear like amputations, or like missing segments of the limb. Longitudinal defects are missing rays of the limb (for example, a missing tibia and great toe).

Reduction defects of the upper limbs The congenital absence of a portion of the upper limb. There are two general types of defect, transverse and longitudinal. Transverse defects appear like amputations, or like missing segments of the limb. Longitudinal defects are missing rays of the limb (for example, a missing radius and thumb).

Renal agenesis or dysgenesis The failure, or deviation, of embryonic development of the kidney.

Spina bifida A neural tube defect resulting from failure of the spinal neural tube to close. The spinal cord and/or meninges may or may not protrude. This usually results in damage to the spinal cord with paralysis of the involved limbs. Includes myelomeningocele (involving both spinal cord and meninges) and meningocele (involving

just the meninges).

Stenosis A narrowing or constriction of the diameter of a bodily passage or orifice.

Stenosis or atresia of large intestine, rectum and anus The absence, closure or constriction of the large intestine, rectum or anus. Can be surgically corrected or bypassed.

Stenosis or atresia of the small intestine A narrowing or incomplete formation of the small intestine obstructing movement of food through the digestive tract.

Tetralogy of Fallot A congenital cardiac anomaly consisting of four defects: ventricular septaldefect, pulmonary valve stenosis or atresia, displacement of the aorta to the right, and hypertrophy of right ventricle. The condition is corrected surgically.

Tracheoesophageal fistula An abnormal passage between the esophagus and trachea. Leads to pneumonia. Corrected surgically. It is frequently associated with esophageal atresia.

Translocation The rearrangement of genetic material within the same chromosome or the transfer of a segment of one chromosome to another one. People with balanced translocations do not always manifest genetic syndromes, but may be carriers of genetic syndromes and can have children with unbalanced translocations. Can occur with any chromosomal anomaly syndrome.

Transposition of the great vessels A congenital malformation in which the aorta arises from the right ventricle and the pulmonary artery from the left ventricle (opposite of normal), so that the venous return from the peripheral circulation is recirculated without being oxygenated in the lungs. Immediate surgical correction is needed. When this is not associated with other cardiac defects, and not corrected, it is fatal.

Tricuspid valve atresia or stenosis A congenital cardiac condition characterized by the absence or constriction of the tricuspid valve. The opening between the right atrium and right ventricle is absent or restricted, and normal circulation is not possible. This condition is often associated with other cardiac defects. This condition is surgically corrected depending on the severity.

Trisomy A chromosomal abnormality characterized by one more than the normal number of chromosomes. Normally, cells contain two of each chromosome. In trisomy, cells contain three copies of a specific chromosome.

Trisomy 13 (Patau syndrome) The chromosomal abnormality caused by an extra chromosome 13. The extra copy can be free-lying, or can be attached to some other chromosome. Trisomy 13 can occur in mosaic so that there is a population of normal cells and a population of trisomy 13 cells. Trisomy 13 is characterized by impaired midline facial development, cleft lip and palate, polydactyly, and mental retardation. Most infants do not survive beyond 6 months of life.

Trisomy 18 (Edwards Syndrome) The chromosomal abnormality characterized by an extra copy of chromosome 18. The extra chromosome can be free lying or attached to another chromosome. Trisomy 18 can occur in mosaic so that there is a population of normal cells and a population of trisomy 18 cells. Trisomy 18 is characterized by mental retardation, neonatal hepatitis, low-set ears, skull malformation, and short digits. Cardiac and renal anomalies are also common. Survival for more than a few months is rare.

Trisomy 21 (Down Syndrome) The chromosomal abnormality characterized by an extra copy of chromosome 21. In rare cases this syndrome is caused by translocation. The extra copy can be free-lying, or can be attached to some other chromosome, most frequently number 14. Trisomy 21 can occur in mosaic, so that there is a population of normal cells and a population of trisomy 21 cells. Trisomy 21 is characterized by moderate to severe mental retardation, sloping forehead, small ear canals, flat bridged nose, and short fingers and toes. One third of infants have congenital heart disease, and one third have duodenal atresia. (Both can be present in the same infant.) Affected people can survive to middle or old age. There is an increased incidence of Alzheimer disease in adults with Trisomy 21.

Truncus arteriosus *See Common truncus*.

Ventricle One of the two lower chambers of the heart (plural ventricles). The right ventricle sends blood to the lungs, and the left ventricle passes oxygen-rich blood to the rest of the body.

Ventricular septal defect (VSD) A congenital cardiac malformation in which there are one or several openings in the ventricular septum (muscular and fibrous wall between the right and left ventricle or right and left lower chambers of the heart) allowing a mixing of oxygenated and unoxygenated blood. The openings vary in size and may resolve without treatment or require surgical treatment.